

AMERICAN BOARD OF MEDICAL GENETICS

Logbook Guidelines for Certification in Medical Biochemical Genetics *For Experienced certified Clinical Geneticists*

Purpose: The purpose of the logbook is to document that the applicant has had direct and meaningful involvement in the examination, diagnosis, communication of information, and management of biochemical genetics patients. The logbook cases must provide evidence of well-rounded experience with a wide variety of biochemical genetic conditions. Disorders submitted in the logbook should represent the broad spectrum of prenatal, pediatric, and adult biochemical genetics.

Submission Requirements: Logbooks must be completed in accordance with the written instructions provided in this document.

Case Selection:

1. The Board requires that 150 cases be obtained during the 2 years prior to submission of this application. No more than 5 cases can be obtained in any one day.
2. The Board *requires* that no more than half of the cases be obtained in any four-month period and prohibits the attainment of more than 35 cases in any one-month period.
3. Each logbook entry must document the applicant's role(s) in patient care, including examination, diagnosis and interpretation, communication of the test results, and management.
4. A given patient or family may appear at most three times (initial diagnosis, management in inpatient or emergency room setting, outpatient visit) in an individual's logbook.

Description of Logbook Headings/Columns:

- **Entry number:** Cases are electronically numbered from 1 to 150. You 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 used anywhere in the logbook. Logbooks that contain specific information regarding the identity of a patient will not be reviewed.
- **Date:** The date in month/day/year [MM/DD/YYYY] format identifies when the patient was seen. All cases must be listed in chronological order.
- **Primary Medical Biochemical Genetics Category:** For each case, use the numbers 1 through 5 as outlined below to identify the single category that best describes the indication for seeing the patient. Observe limits per category where specified.

Category 1 **Diagnostic evaluation, new patient (outpatient):** Evaluation of infants, children, or adults initiated because of clinical signs, symptoms, positive family history or abnormal

laboratory results (including newborn screening) suggesting a biochemical genetic condition. The Board requires that at least 25 cases be obtained in this category. Of these, no fewer than 5 and no more than 15 should involve newborn screening.

Category 2 **Diagnostic evaluation, new patient (inpatient):** Evaluation of infants, children, or adults initiated because of clinical signs, symptoms, or abnormal laboratory results (including newborn screening) in the inpatient setting. The Board requires that at least 10 cases be obtained in this category.

Category 3 **Prenatal diagnosis:** Counseling patients and families regarding the availability, risks and implications of prenatal testing for metabolic disease, coordinating prenatal testing, and/or interpreting and communicating prenatal test results to patients and families. Discussion of potential prenatal testing and its implications can be included in this category. It is *recommended* that at least 10 cases be obtained in this category.

Category 4 **Management/continuing care, routine (outpatient):** Medical management, consultation, or evaluation of patients previously diagnosed with a biochemical genetic condition. The Board requires that at least 75 cases be obtained in this category.

Category 5 **Management/continuing care, emergent (inpatient or emergency room):** Medical management, consultation, or evaluation of patients previously diagnosed with a biochemical genetic condition in the inpatient setting. The Board requires that at least 5 cases be obtained in this category.

- **Diagnoses:** The logbook cases must demonstrate experience with a variety of medical biochemical genetics cases. The Board *requires* that no more than 40 cases be any one diagnosis, such as phenylketonuria or galactosemia. Variations of specific diagnoses (e.g., phenylketonuria and persistent hyperphenylalaninemia) may not be considered sufficient to count them as separate diagnoses, and replacement cases may be requested when the aggregate exceeds the case limit. A maximum of 40 cases may be those evaluated for a metabolic disorder but determined to be normal.

Follow the instructions below when entering diagnoses in the online logbook:

1. Record the primary diagnosis for each case, using consistent and standard terminology to describe the biochemical genetic condition. Non-standard abbreviations for the names of the disorders are not acceptable.
2. For cases where no disorder is identified, list the primary reason for metabolic evaluation (e.g., cardiomyopathy, liver disease).
3. For evaluations initiated because of family history or at-risk pregnancy, list the diagnosis first followed by the justification for classification in this category (e.g., OTC deficiency, positive family history).
4. If more than one patient or family with the same biochemical genetic category, diagnosis, visit date, trainee role(s), and supervisor are recorded, clearly indicate that entries are not duplicated records or members of the same family, as follows: “Phenylketonuria, patient or family 1; Phenylketonuria, patient or family 2”.

- **Experienced Clinical Geneticist's Roles:** Check all of the boxes that indicate the nature of your involvement in the case. A breadth of experience must be reflected in the logbook. A minimum of three roles must be specified for at least 120 cases.
 1. **Patient encounter:** Collection and review of records, medical history, family history and physical examination. At least 45 cases must include this category of involvement.
 2. **Evaluation plan/Diagnosis/Interpretation of results (new patients):** Pertinent background reading, evaluation of the clinical significance of findings, generating a differential diagnosis, recommending further testing, and summarizing the case. At least 20 cases must involve this role.
 3. **Ongoing management (follow-up patients):** Follow the disease progression with appropriate tests and medical management; provide appropriate counseling, psychosocial support, and anticipatory guidance to the family. At least 85 cases must include this category of involvement.
 4. **Communication of results to other professionals:** Provide an oral and/or written discussion of results and their significance to the referring service or ancillary services. At least 30 cases must involve this role.
 5. **Oral communication of results to patients:** Provide an oral and/or written discussion of results and their significance to patients and families, including, as appropriate, results and implications of clinical examination, laboratory tests, procedures differential diagnosis and management options. At least 60 cases must include this category of involvement.
 6. **Laboratory testing:** Direct involvement in the clinical biochemical genetics laboratory, including interpretation of primary laboratory data, generating a differential diagnosis, and making a plan for evaluation of the patient. Data may be obtained from a laboratory not onsite. The Board *requires* that at least 15 cases involve this role.