Certification in Clinical Genetics and Genomics

Logbook Requirements for

2026 Examination

Purpose:

The purpose of the logbook is to document that the applicant has had direct and meaningful involvement in the genetic evaluation, counseling, and management of patients and/or families, and has received appropriate clinical supervision. The logbook submitted must provide evidence of a well-rounded experience in all clinical genetic categories involving a range of roles in evaluation, risk assessment, genetic counseling, genetic testing and results provision, and management. Clinical categories and genetic diagnoses should include the broad spectrum of prenatal, pediatric, and adult genetics with a wide variety of diagnoses. The diagnostic categories should include, but are not limited to, developmental delays and intellectual impairment, chromosomal disorders, multiple malformation syndromes, metabolic diseases, prenatally diagnosed abnormalities, neurological disorders, common complex disorders, and cancer. Applicants should also have experience with consenting families and delivering testing results for exome sequencing and panel testing and with inpatient and outpatient encounters.

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 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 and 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 cases must be obtained through an ACGME-accredited medical genetics and genomics residency.

2. Supervision for patient encounters in genetics clinics must be provided by faculty certified by the ABMGG, ABGC, or CCMG. For cases obtained during rotations in specialty clinics, e.g., oncology, neurology, it is *recommended* that supervisors be certified by their appropriate ABMS certifying board(s). All supervisors should be identified in the training program's accreditation documents.

3. All 150 cases must be obtained during the inclusive dates of the applicant's medical genetics and genomics residency training. No more than 5 cases may be obtained in any one day.

4. Each logbook entry must document a direct interaction between the resident and an individual patient and/or family. Telemedicine cases may be included in the logbook as long as the appropriate learning objectives have been fulfilled. A minimum of 75% of cases must be in-person, face-to-face encounters. A minimum of 10% of telemedicine cases is recommended.

5. A given patient or family may appear only once in an applicant's logbook, regardless of the number of encounters with that patient or family.

6. No more than 30 cases may have a genetic counselor listed as the primary supervisor.

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 of 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 not be included. 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 when the patient was evaluated.

- **Patient Age Category: For** each case, the patient's age must be defined as Infant, Child, Adolescent, or Adult. Age refers to the age of the patient on the date of the clinical visit. Note: in Primary Genetic Category 3 (next section), the pregnant woman (or adolescent) is the patient, so Adult or Adolescent are the only appropriate options.

- **Primary Genetic Category:** For each case, use the numbers 1 through 4, as outlined below, to identify the category that best describes the indication for the clinic visit. A minimum of 15 cases and a maximum of 70 cases must be documented in each category.

  Category 1 **Diagnostic evaluation:** Evaluation of infants, children, or adults with signs or symptoms of a genetic condition.

  Note: Individuals with signs or symptoms of a genetic disorder, including those with a personal history of cancer seeking genetic evaluation/risk assessment and infants who were found to have true or false positive newborn screening tests, should be logged in this category.

  Examples: Congenital anomalies, intellectual disability/developmental delay, inborn errors of metabolism suspected by symptoms or newborn screening, Mendelian disorders, common complex disorders, cancer, and cytogenetic abnormalities.

Category 2 **Genetic risk assessment:** Evaluation of asymptomatic or presymptomatic individuals with known or suspected risk factors for a genetic condition, including family history. Counseling regarding genetic testing and genetic testing results provision.

Note: Individuals with a known genetic disorder who are seen for routine medical surveillance for potential sequelae of that disorder, e.g., Turner syndrome for aortic dilation, should not be logged in this category but instead in Category 4.

Examples: Asymptomatic or presymptomatic individuals at risk for carrying an autosomal, X-linked, or mitochondrial gene pathogenic variant because of the following: (a) recurrent reproductive loss or familial chromosome rearrangements, such as balanced translocations; (b) a family history of cancer; (c) a family history of adult-onset neurological disorders; and (d) a family history of other genetic conditions or common complex disorders.

Category 3

Prenatal genetics: Assessment of fetal genetic risk in a current pregnancy with or without prenatal testing.

Examples: Fetal risk assessment for aneuploidy, abnormal prenatal test results such as fetal abnormalities on ultrasound examination, fetal molecular testing or abnormal maternal serum screens, and teratogen exposures. The term "advanced maternal age" as a diagnosis should be avoided. It is *recommended* that abnormal serum screening or fetal risk assessment for aneuploidy comprises no more than 5 cases.

Category 4

Management/continuing care: Medical management or additional consultation or evaluation of individuals with a known genetic diagnosis.

Examples: Inborn errors of metabolism, multiple congenital anomalies, cytogenetic abnormalities, hereditary cancers, neurological disorders, intellectual impairment/ developmental delay, connective tissue disorders, and skeletal dysplasias.

- **Diagnosis: Within** any given category, no more than 5 cases may have the same specific diagnosis. Variations in genotype or phenotype of a specific diagnosis, such as age of onset or particular pathogenic variant, are not considered sufficient to count as separate diagnoses. A minimum of 10 cancer cases is *required*, with no more than 5 of a specific type of cancer. Additionally, a minimum of 10 cases of evaluation for inborn errors of metabolism, which can include newborn screening follow-up, is *required*.

  It is *required* that at least 10 cases in Category 2 and at least 15 cases distributed between Categories 1 and 4 pertain to conditions whose clinical ONSET is typically in adolescence or adulthood. Examples include Huntington disease, Friedreich ataxia, adult Pompe disease, myotonic dystrophy, hereditary breast and ovarian cancer, Lynch syndrome, FXTAS, schizophrenia. An adolescent or adult patient whose condition was present at birth (e.g., PKU)

or first manifested soon after birth (e.g., Prader-Willi syndrome) would not qualify as one of the 15 required cases in Categories 1 and 4 with disease onset in adolescence or adulthood. **It is the age at onset and not the age of diagnosis or the age at which the trainee saw the patient that should be taken into account in satisfying this requirement.**

In addition to the numerical requirement, logbooks will be evaluated for overall case diversity, complexity of case experience, and distribution of diagnoses within categories. For example, a logbook recording the minimum number of 15 cases in Category 3 must have a diverse case set of fetal aneuploidy screening, teratogenesis, maternal genetic conditions in pregnancy and fetal malformations or other conditions. Similarly, review is based on variety in late childhood or adult-onset disorder, i.e., not all cases should be cancer related.

For each case, enter the diagnosis using the guidelines below.

1. Enter the diagnosis using the OMIM name or an OMIM alternative title. All cases representing the same condition should be entered using the same diagnosis name. For example: Enter all PKU cases as "PKU," not "PKU" for some and "phenylketonuria" for others.

2. Do not use abbreviations unless it is an OMIM alternative title. For example, do not substitute MCA for multiple congenital anomalies.

3. Primary diagnosis must be listed first. For example, "Breast cancer, positive family history" *not* "Family history of breast cancer."

4. Use the most specific diagnosis for each case when known, e.g., Fragile X syndrome, or a broader designation when the specific diagnosis is unknown, e.g., intellectual disability, unknown cause.

5. Log only those cases for which the diagnostic evaluation is complete. For example, "5p deletion syndrome" not "Rule out chromosome anomaly." If making a specific diagnosis was the reason for the referral, for example, "is this Marfan syndrome?," use "Marfan syndrome" if the diagnostic evaluation is complete and this is the diagnosis or "Marfan syndrome, excluded" if the diagnostic evaluation is complete and this diagnosis was excluded but a more specific diagnosis could not be made. If a more specific diagnosis could be made, such as Shprintzen-Goldberg syndrome, use the more specific diagnosis.

6. If more than one patient or family with the same genetic category, age 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: Neurofibromatosis, patient or family 1; Neurofibromatosis, patient or family 2.

7. For Primary Genetic Category 2 patients, list not only the diagnosis but also the reason for classification in this primary genetic category. For example: enter "Trisomy 18, previously affected child" *not just* "Trisomy 18." A diagnosis without a justification will not satisfy the requirement, and it will be assumed that the case entry is a patient affected with the condition.

- **<u>Trainee's Roles</u>:** Check all of the boxes that indicate your role(s) in the clinical evaluation, counseling, and management for each case. A minimum of 5 roles must be specified for each case. At least 10 cases ***must*** be obtained for each role; however, it is *recommended* that at least 15 cases be obtained for each role.

1. **Medical history** involves obtaining pertinent medical information, such as pregnancy history, developmental milestones, and environmental exposures, by patient interview and review of medical records.

2. **Pedigree** includes eliciting information for the construction of a three-generation pedigree that includes at a minimum all first-, second- and third-degree relatives using standard symbols.

3. **Physical examination**s entails performing a complete physical examination or, if more appropriate, a targeted examination, to assess the system(s) of concern or to look for manifestations of a Mendelian condition in individuals who present for evaluation of a common complex disorder.

4. **Management/Evaluation plan** involves determining recommendations for appropriate tests and/or assessments of medical or psychosocial care for a patient/family.

5. **Testing options** includes explaining the technical and medical aspects of diagnostic and screening methods and reproductive options, including associated risks, benefits, and limitations of testing.

6. **Testing results** includes variant interpretation and communicating testing results.

7. **Risk assessment** entails performing pedigree analysis and evaluation of medical and laboratory data to determine recurrence/occurrence risks.

8. **Inheritance/risk counseling** involves educating the patient or family about recurrence/occurrence risks and modes of inheritance of the disorder.

9. **Discussion of diagnosis/natural history** includes conveying genetic medical information about the diagnosis, etiology, natural history, prognosis, and treatment/management of the disorder(s) in question.

10. **Psychosocial support/counseling** involves providing short-term, patient or family-centered counseling, psychosocial support, and anticipatory guidance to the family, as well as addressing patient concerns.

11. **Information access** includes literature review and database searches, as well as identification of resources for the patient or family and referring healthcare provider.

12. **Documentation and follow-up** involve writing a consultation report or letter to the family or healthcare provider and recording adequate follow-up notes.

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