

Recognition of Board-Certified Clinical Laboratory Professionals as Qualified Healthcare Professionals

As laboratory testing increases in complexity, doctoral level (PhD or MD/DO) diagnostic laboratory professionals, such as clinical laboratory geneticists, are expected to provide complex laboratory interpretation in the context of a patient's medical history, family history, and/or other test results.^{1,2} For example, clinical genetic diagnostic laboratories provide detailed reports to the ordering physicians, which include an individualized clinical interpretation of the results including clinical implications, significance of the findings, and when applicable, recommendations for follow-up testing and genetic counseling. For accreditation of a genetics laboratory, the College of American Pathology (CAP) specifies that the report must include "*a discussion of the limitations of the findings and the clinical implications of the detected mutation (or negative result) for complex disorders with regard to recessive or dominant inheritance, recurrence risk, penetrance, severity and other aspects of genotype-phenotype correlation.*"³ These reports also must be clearly written and understandable to non-geneticist professionals.

While the technical components are covered under the clinical laboratory fee schedule (CLFS), clinical test interpretation for most of these tests is covered under the physician fee schedule (PFS), and only qualified healthcare professionals (QHPs) can directly bill Medicare under the PFS. However, board-certified doctoral level laboratory professionals (including PhDs and MD/DO trained individuals not licensed to practice medicine in the US) currently do not qualify as QHPs and thus cannot bill Medicare directly for interpretive services. For doctoral-level clinical laboratory professionals to be recognized as QHPs, legislation is needed. Numerous other types of non-physician clinical professionals have already successfully achieved recognition as QHPs (*e.g.*, clinical psychologists, physical therapists, physician assistants), and legislative initiatives for many others are currently underway (*e.g.*, genetic counselors).

Recommendation: Appropriately trained and board-certified doctoral level diagnostic laboratory professionals should be recognized by Medicare as QHPs for the purpose of *individualized clinical interpretation of laboratory findings* provided 1) they possess a doctoral degree in a relevant life sciences discipline; 2) their training and certification include certain **key components** (outlined below) designed to support such individualized clinical interpretation; and 3) they review and combine analytical results, clinical findings,

¹ Aziz N, Zhao Q, Bry L, et al. College of American Pathologists' laboratory standards for next-generation sequencing clinical tests. *Arch Pathol Lab Med*, 2015 Apr; 139(4):481-93

² Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology*, 2015 May; 17(5):405-24

³ College of American Pathologists, Master Molecular Pathology Checklist. 09/25/2012; <http://webapps.cap.org/apps/docs/education/OnlineCourseContent/2012/LAP/Resources/Checklists/MOL09252012.pdf>

other laboratory results, and family history when appropriate to issue interpretive reports in a high complexity CLIA-certified laboratory.

Individualized clinical interpretation of laboratory findings includes generation of an individualized report that considers analytical findings within the context of clinical indications such as patient and family medical history, clinical phenotype, and other laboratory results. The report is provided to the ordering health care provider to facilitate patient diagnosis, management, and treatment, including recommendations for additional testing when necessary. It should be recognized that *individualized clinical interpretation* goes beyond the standard technical analysis of results and classification of analytical findings. Development of an *individualized clinical interpretation* requires communication with the ordering clinician to obtain and review relevant medical and/or family history information. The clinical laboratory report containing the individual clinical interpretation performed by the board-certified laboratory professional is a critical part of the medical record and provides the content and context of laboratory findings to aid the patient's health care providers with an accurate diagnosis, prognosis, management, and treatment of disease when combined with the medical evaluation and other clinical findings.

Examples of situations that generally require an individualized clinical interpretation:

- 1) An example of an individualized interpretation occurs as part of the analysis of a clinical exome or genome. This requires not only molecular expertise to identify potentially pathogenic variants, but also expertise in medical genetics to determine whether the identified variant is clinically relevant and should be reported. This expertise depends upon skills beyond applying defined criteria to classify a variant. Clinical laboratory reports generated from exome and genome sequencing provide guidance to the patient's clinician(s) for management, counseling, and identification of additional family members who may benefit from testing.
- 2) A second example of this type of individualized interpretation is the analysis and reporting of complex karyotypes by cytogeneticists in patients with hematologic malignancies. This interpretation integrates findings from morphologic examination of the marrow and flow cytometry along with other clinical features to place the cytogenetic findings into the correct clinical context for the patient. Cytogeneticists may also recommend additional molecular testing that could augment the diagnostic and prognostic value of their results.
- 3) A third example is the interpretation of organic acid analysis, which requires integration of knowledge about specific disorders, the pattern of metabolites detected and their relative significance, and the review of other clinical and laboratory data that could support or detract from a specific diagnosis and recommendations for further testing.

Examples of situations that generally do not require an individualized clinical interpretation:

- 1) Targeted variant assays, such as factor V Leiden or HFE-associated hereditary hemochromatosis for which results can generally be released without knowledge of symptoms, family history or other laboratory results, and reports can utilize a pre-formatted template.
- 2) The reporting of a normal karyotype or a relatively common aneuploidy such as trisomy 21 in a patient being screened for constitutional chromosomal abnormalities or a targeted fluorescence in situ hybridization (FISH) assay designed to monitor patients with a history of the targeted gene fusion (*e.g.* detection of *BCR/ABL1* fusion in patients with CML).
- 3) Measurement of hexosaminidase enzyme activity for the diagnosis of Tay-Sachs disease

Required Key Components of Training and Board-Certification:

- Broad technical and analytical training to support integration of other laboratory findings (beyond the field of the specialized training);
- Clinical fellowship training/rotations directly with patients designed to support interpretation of laboratory findings within the context of medical evaluation findings, phenotype, and family history;
- Specialized training to support preparation and generation of reports that incorporate all relevant clinical and laboratory data, are designed to facilitate diagnosis, management and treatment of disease, and are written such that information is clearly understood by the ordering health care provider;
- A program for continuing certification that is designed to ensure that diplomates are engaged in career-long learning and remain current in their specialty training;
- Training consistent with the type of services the laboratory professional is interpreting (*e.g.*, infectious agents, HLA typing, inherited and somatic variation); and
- Achievement of a passing score on a comprehensive certification examination given by the relevant certification board.

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