



Health Level Seven® International

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For Immediate Release

Health Level Seven Clinical Genomics Version 2 Messaging Standard Implementation Guide Successfully Transmits Genomic Data Electronically

The Partners HealthCare Center for Personalized Genetic Medicine and Intermountain Healthcare Clinical Genetics Institute successfully piloted the implementation guide, which contains a structured and coded data model for transmitting genetic test results (and associated interpretations), from the testing laboratory into the patient's electronic health record (EHR).

Ann Arbor, Michigan, USA – January 19, 2010 – Health Level Seven (HL7®), the global authority for interoperability and standards in healthcare information technology, today announced its Version 2 messaging standard has successfully gathered structured and coded genetic tests results from a lab and transmitted them to an individual patient's EHR for the first time. The current standard for genetic test results is either paper or electronic narrative reports.

The guide, titled *HL7 Version 2 Implementation Guide: Clinical Genomics; Fully LOINC-Qualified Genetic Variation Model, Release 1*, details structuring a genetic test result into the EHR utilizing HL7 Version 2.5.1. It is based on both the *HL7 Version 2*

Implementation Guide Laboratory Result Reporting to the EHR, and the HL7 Version 3 Genetic Variation data model. The guide covers the reporting of genetic test results for sequencing and genotyping based tests where identified DNA sequence variants (i.e. mutations) are located within a gene. This includes testing for DNA variants associated with disease and pharmacogenomic applications, such as predicting a patient's responsiveness to drug therapy and drug metabolism rate. It is fully-LOINC qualified, meaning that new LOINC codes have been created to represent the test components and results. LOINC codes offer the consistency of representation across different message types and for clinical decision support.

The implementation guide was used by The Partners HealthCare Center for Personalized Genetic Medicine (PCPGM) and the Intermountain Healthcare Clinical Genetics Institute to gather genetic test results and transmit them to an individual patient's electronic health record for the first time. The results were sent by a direct computer interface from PCPGM to Intermountain Healthcare.

"This work aligns with national efforts to re-examine and improve healthcare delivery," says Mollie Ullman-Cullere, PCPGM senior information architect and co-chair of HL7 Clinical Genomics Work Group. The work group is comprised of volunteers who come from prominent healthcare systems, major laboratories, and leading health IT software vendors.

"The project is among the first in the country that will create a standardized advanced electronic patient record system containing genetic data," says Stan Huff, chief medical informatics officer for Intermountain Healthcare and HL7 International board member. "This may lead to the electronic health record of the future, which would support treatment plans that are tailor made for each individual — right down to their DNA."

Huff's team at Intermountain Healthcare worked with Partners HealthCare for 14 months to build the framework for receiving test results and integrating them into a patient's electronic health record. During this time the Partners team developed a lab reporting system that would create and send out the test result message through a

centralized interface hub. Any lab or EHR that implements this HL7 standard can interface with this hub.

Utilizing this Version 2 implementation guide, Intermountain and Partners Healthcare are working to make this genetic information available within the EHR, including clinical decision support, linkage to clinical genetic knowledge bases (keeping clinical interpretations on the variants up-to-date), and tools for pharmacogenomics and drug order entry. Clinicians expect to use genetic data for confirmatory diagnosis or risk for developing the disease, and determination of drug metabolism, drug efficacy, or drug toxicity.

Forming a more complete diagnostic picture for inherited conditions may require augmenting genetic data with family history data, represented by the HL7 Version 3 Pedigree model. This is especially important, because genetics isn't about just treating the patient, but treating the whole family.

"Now we can begin to write programs and protocols that will take this information and suggest to clinicians the best personalized health care for each individual, based on their genes," says Huff. "A physician will not need to be an expert in family medicine *and* genetics — our programs will help."

HL7 International members may download a copy of the implementation guide for free. Nonmembers may purchase the guide from the HL7 store at <http://www.hl7.org/store/index.cfm>

About Health Level Seven International (HL7)

Founded in 1987, Health Level Seven International is the leading global authority for healthcare Information interoperability and standards with affiliates established in more than 30 countries. HL7 is a non-profit, ANSI accredited standards development organization dedicated to providing a comprehensive framework and related standards for the exchange, integration, sharing, and retrieval of electronic health information that supports clinical practice and the management, delivery and evaluation of health

services. HL7's more than 2,300 members represent approximately 500 corporate members, which include more than 90 percent of the information systems vendors serving healthcare. HL7 collaborates with other standards developers and provider, payer, philanthropic and government agencies at the highest levels to ensure the development of comprehensive and reliable standards and successful interoperability efforts.

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