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Stelios G. Sfakianakis received his BSc in Computer Science in 1995 and his MSc with highest distinction in advanced information systems in 1998 from the University of Athens, Greece. Since January 2000 he works at the Biomedical Informatics Laboratory of the Institute of Computer Science, Foundation for Research and Technology – Hellas (FORTH) in Heraklion, Greece. His work is focused on the design and implementation of a component based architecture for the Integrated Electronic Health Record using CORBA and Web Services as the middleware technology. His interests and work span from the design of relational or hierarchical databases to visual modeling with UML, open source operating systems, and distributed object and service oriented computing.

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Catherine E. Chronaki, BEng (88), MSc (90) Catherine is a senior software engineer with FORTH-ICS, biomedical laboratory managing the internal group on eHealth platforms. Since 1992, she has been involved in a number of health-related national and EU funded projects. Over the years, Catherine has contributed to the design, implementation, and evaluation of a range of eHealth systems and services from I2C, a system for the indexing and retrieval of medical images by content, to medical collaboration environments and middleware supporting the Integrated EHR in regional health information networks. Currently, she is the technical manager at FORTH-ICS for the Twister project investigating eHealth services over hybrid wireless-satellite networks in Rural areas and the eHealth Consumer trends survey that measures consumer attitude towards eHealth. She is also the coordinator of OpenECG (www.openecg.net), a network with world-wide impact promoting interoperability standards for electrocardiography. Recently, she has coordinated an HL7-Hellas SIG on technical implementation guidelines for HL7. Catherine's interests are in the area of biomedical engineering, and eHealth adoption. She interested in EHR developments, medical databases, interoperability of medical devices, Internet services, and she is intrigued by the notion of individualized health monitoring.. Catherine has authored or coauthored more than 40 scientific publications.

Title

Early experience from HL7 v3 tools, the Pedigree topic, and CDA in the Danish HNPCC Registry

Abstract Covers

case study

Suggested length of presentation

20 minutes

Description

Genetic departments, laboratories, and surgical departments gather clinical and genomic information about an individual and submit this information to a Hereditary Non Polyposis Colon Cancer (HNPCC) registry along with a family identification number. The objective of this study is to investigate the appropriateness of the HL7 v3 family of standards in delivering clinical and genomic data to the Danish HNPCC Registry in a solution that conforms to international standards. This is the first step in linking HNPCC registries throughout the world for the benefit of personal health care and clinico-genomic research.

Abstract

Early experience from HL7 v3 tools, the Pedigree topic, and CDA in the Danish HNPCC Registry

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Hereditary Non Polyposis Colon Cancer (HNPCC) registries strife to improve the prognosis and quality of life for HNPCC families by collecting and integrating demographic and disease-related data on family members for family identification, counseling, surveillance and research to establish diagnosis and risk calculation in the presence of ever increasing data and research results. INFOBIOMED (http://www.infobiomed.org), a European-funded Network of Excellence, aims to enable systematic progress in clinical and genetic data interoperability and integration, while at the same time advancing the exchange and interfacing of methods, tools, and technologies used. This study reports early experience for a currently running INFOBIOMED pilot that employs the Danish HNPCC Registry as a model to demonstrate the synergistic effect of combining clinical and genomic information for families with increased risk of a genetic disease, using HL7 v3.

Currently in Denmark, genetic departments, laboratories, and surgical departments are the main entities reporting to the HNPCC registry. All genetic departments involved in the pilot use a local registration system and Cyrillic is their typical pedigree program. Laboratories do not use a

registration system; therefore, HL7-based communications of molecular based information would assist the standardized collection of epidemiological information from different research groups. Surgical departments transmit electronically in EDIFACT discharge letters to the family doctor. Nevertheless, none of the four surgical departments involved in the pilot has an Electronic Health Record system for registering the endoscopies performed and reporting to the national HNPCC-Registry is in hard copy. Despite the inherent heterogeneity of data sets and sources in departments that involve the treatment of HNPCC families, all departments expressed willingness to participate in the development of harmonized reporting on HNPCC. As a result, a set of XML schemas were developed and in this study we explore the mapping of these schemas into HL7 v3 messages and report on our experience and the main lessons learned.

The relevant area of work within HL7 is the Pedigree Topic (Family History) of the Clinical Genomics SIG (ref.1), which aims "to illustrate the way a patient's pedigree with clinical and genomic data could be represented for risk analysis purposes in the context of breast and ovarian cancers and other diseases". The Pedigree model clearly defines the two application roles that we have in our case: the Pedigree Sender, which can be a surgical department or any other clinical and genomic information source, and the Pedigree Receiver, which is the HNPCC Registry. However, the problem identified here was the inability of the Pedigree Model to represent a general «clinical statement» for a person. Instead, the HL7 model supports a single Clinical Observation that in future versions will be replaced by the HL7 Clinical Statement model. This was a major issue for us since the information submitted by the various information sources to the HNPCC Registry usually has a rich content structure as reflected by the preexisting XML schemas. Although this content structure can be modeled with a clinical statement as included in CDA release 2, it could not be represented as in the Pedigree Topic with a single clinical observation.

The Pedigree Model, however, seemed very close to the desired solution so we thought it would be a good idea to create a localization profile that supports the rich content structure of the report messages sent to the HNPCC Registry. The HL7 Tools (ref.2) (RMIM Designer and HL7 v3 Generator) were employed to enhance the Pedigree Model with the ability to compose and relate Clinical Observations. However the tools did not work the way we assumed! This is not a surprise since extending an HL7 model by adding a new Act Relationship does not conform to the HL7 guidelines as described in «Refinement, Constraint and Localization» document (ref.3). Major limitation in this approach was that the XML schemas produced would be in the HL7 namespace and would use the naming conventions of HL7, leading to conflicts and ambiguities if used jointly with other HL7 conformant parties.

Finally, we looked into the Clinical Document Architecture (CDA) Release 2 (ref.4), which incorporates a clinical statement that seems to fit our purpose. Although the CDA standard is not specific for pedigree or genomic information, it seems to be a tool general enough for the realization of HNPCC use cases. The CDA approach is currently being implemented with no major issues so far. However, the adoption of CDA requires much discipline both from the design and the implementation viewpoint. For example, the use of vocabularies (either local or international/standard ones) is enforced everywhere and attention should be paid to the proper coding of the information.

The presence of multiple HNPCC registries in Europe and throughout the world operating under different technology, methods, tools, and infrastructure, signifies a pressing need for robust HL7 standards enabling the much needed interoperability. Based on experience in this study we conclude that following the CDA approach is currently the preferred way of implementing the HNPCC Registry interaction scenarios. Nevertheless, as the Family History work of the Clinical Genomics SIG incorporates more feedback from implementation, it will become more expressive and achieving interoperability among national HNPCC registries will be a simpler task for the benefit of personal health care and clinico-genomic research.

References

(ref.1) HL7 Clinical Genomics, Version 3 Standard http://www.hl7.org/v3ballot/html/domains/cg/hmpocg.htm

(ref.2) HL7 Tools http://www.hl7.org/Library/data-model/V3Tooling/toolsIndex.htm

(ref.3) HL7 «Refinement, Constraint and Localization", Version 3, Release 2, Committee 3 Ballot
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(ref.4) HL7 Clinical Document Architecture, Release 2.0 ANSI/HL7 CDA, R2-2005
4/21/2005 http://www.hl7.org/v3ballot/html/infrastructure/cda/cda.htm