Family History
Information Exchange
Services Using HL7 Clinical Genomics Standard Specifications

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ABSTRACT

A number of family history applications are in use by health care professionals (e.g., CAGENE, Progeny, Partners Health care Family History Program) as well as by patients (e.g., the US Surgeon General’s Family History Program). Each has its own proprietary data format for pedigree drawing and for the maintenance of family history health information. Interoperability between applications is essentially non-existent. To date, disparate family history applications cannot easily exchange patient information. The receiving application should be able to understand the semantics of the incoming family history and enable the user to view and/or to edit it using the receiving applications interface. We envision that any family history application will be able to send and receive an individual’s family history information using the newly created HL7 Clinical Genomics Specifications through the Semantic Web, using services that will transform one format to the other through the HL7 canonical representation.

Keywords: breast cancer risk assessment; clinical genomics; electronic health record; family history; health informatics standards

INTRODUCTION

The need to represent a patient’s family history information associated with clinical and genomic data was introduced as a storyboard to the Clinical Genomics Special Interest Group (SIG) in HL7 (CG, 2005). The SIG develops HL7 standards (HL7, 2005) to enable the exchange of interrelated clinical and personalized ge-
nomic data between disparate organizations (e.g., health care providers, genetic labs, research facilities, pharmaceutical companies). Agreed-upon standards to allow this exchange are crucial, as it is envisioned that the use of genomic data in health care practice will become ubiquitous in the near future. A few emerging cases for this include tissue typing, genetic testing (e.g., cystic fibrosis, BRCA1, BRCA2), and pharmacogenomics clinical trials. These cases are represented in the SIG storyboards, which have led to the development of the Genotype model as the basic unit of genomic data representation, focusing on a specific chromosomal locus.

It was determined that there was a set of basic information required to record a family history and to create a pedigree for the purpose of breast cancer risk assessment (Thull & Vogel, 2004). For each family member, this set included the information about his or her relationships to other members of the family and the information regarding his or her health. Relationship information included the type of relative, a personal identifier, and the identifier of the person’s mother and father. Health information data included disease type, age at diagnosis, current age or age of death, genetic syndrome suspected, genetic test done, genetic test result as raw data, and interpretation of genetic test.

The explosion in our knowledge of genetics has increased our understanding of the hereditary basis of many diseases. While we present here the example of exchanging family history and risk information relative to breast cancer, we believe this model can be used for the exchange of any hereditary risk information.

An outline of the patient’s family history is presented in Appendix A. Populating this data set with patient data results is the example shown in Table 1.

**Storyboard Presentation**

The following fictitious scenario demonstrates the potential use of the Semantic Web (Berners-Lee, Hendler & Lassila, 2001) in offering services of exchanging family history information. Note that this is an abridged version of the full presentation contained in the HL7 specifications (CG, 2005).

1. Martha Francis is 39 years old. Her mother had ovarian cancer and was found to have a deleterious BRCA1 mutation. She has two sisters, a husband, and a daughter. She is not of Ashkenazi Jewish descent.
2. She makes an appointment at a risk clinic. The clinic instructs her to use the Surgeon General’s Family History (Yoon, 2002) Web-based tool to prepare for the visit. She brings up the Surgeon General’s Family History tool and enters her family history.
3. She then sends the data to the risk clinic prior to her appointment with that clinic, where a CAGENE application receives the data.
4. The counselor at the risk clinic (nurse geneticist, nurse practitioner, genetic counselor, doctor, etc.) uses the CAGENE application (a pedigree drawing program that runs risk models), where the patient’s family history
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