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Version 1.1

HITSP Family History Decision Support for Genetic Risk Analysis Component

HITSP/C90

Healthcare Information Technology Standards Panel

Submitted to:

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Submitted by:

Care Management and Health Records Domain Technical Committee

DOCUMENT CHANGE HISTORY

Version Number	Description of Change	Name of Author	Date Published
	Template Update to V2.4	Project Team	July 31, 2008
0.0.1	Review Copy	Care Management and Health Records Domain Technical Committee	September 26, 2008
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1.0.1	Review Copy	Care Management and Health Records Domain Technical Committee	June 30, 2009
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FIGURES AND TABLES

1.0 INTRODUCTION

1.1 OVERVIEW

The HITSP Family History Decision Support for Genetic Risk Analysis Component is used to communicate genetic and family history information from healthcare IT applications to a clinical decision support system that provides an assessment of genetic risk of disease for a patient. It uses the HL7 Version 3 Standard to define the content of an HL7V3 message including: Clinical Genomics; Pedigree, Release 1 to support the communication of genetic and family history information to the clinical decision support system, and to support the communication of risk information from that system back to the originator. The specification for the transport of this message content is outside the scope of this construct.

1.2 COPYRIGHT PERMISSIONS

1.3 REFERENCE DOCUMENTS

This section provides a list of key reference documents and background material. If you are already familiar with this information, proceed to Section 2.0.

A list of key reference documents and background material is provided in the table below. These documents can be retrieved from www.hitsp.org.

Table 1-1 Reference Documents

Reference Document	Document Description
HITSP Acronyms List	Lists and defines the acronyms used in this document
HITSP Glossary	Provides definitions for relevant terms used by HITSP documents
TN900 - Security and Privacy	TN900 is a reference document that provides the overall context for use of the HITSP Security and Privacy constructs
TN901 - Clinical Documents	Developed as a reference document to provide the overall context for use of the HITSP Care Management and Health Records constructs

1.4 CONFORMANCE

This section describes the conformance criteria, which are objective statements of requirements that can be used to determine if a specific behavior, function, interface, or code set has been implemented correctly.

1.4.1 CONFORMANCE CRITERIA

In order to claim conformance to this construct specification, an implementation must satisfy all the requirements and mandatory statements listed in this specification, the associated HITSP Interoperability Specification, its associated construct specifications, as well as conformance criteria from the selected base and composite standards. A conformant system must also implement all of the required interfaces within the scope, subset or implementation option that is selected from the associated Interoperability Specification.

Claims of conformance may only be made for the overall HITSP Interoperability Specification or Capability with which this construct is associated.

1.4.2 CONFORMANCE SCOPING, SUBSETTING AND OPTIONS

A HITSP Interoperability Specification must be implemented in its entirety for an implementation to claim conformance to the specification. HITSP may define the permissibility for interface(s) scoping, subsetting or implementation options by which the specification may be implemented in a limited manner. Such scoping, subsetting and options may extend to associated constructs, such as this construct. This construct must implement all requirements within the selected scope, subset or options as defined in the associated Interoperability Specification to claim conformance.

2.0 COMPONENT DEFINITION

2.1 CONTEXT OVERVIEW

The text adapted from HL7 Version 3.0 Clinical Genomics begins here:

> The HL7 Clinical Genomics domain addresses requirements for the interrelation of clinical and genomic data at the individual level. Much of the genomic data are still generic, for example the human genome is in fact the DNA sequences believed to be the common sequences in every human being. The vision of 'personalized medicine' is based on those correlations that make use of personal genomic data such as the SNPs (Single Nucleotide Polymorphisms) that differentiate any two persons and occur about every thousand bases. Beside normal differences, health conditions such as drug sensitivities, allergies and others could be attributed to the individual SNPs or to differences in gene expression and proteomics.
>
> The Clinical Genomics domain is the personalization of the genomic data and the 'intelligent' linking to relevant clinical information. These links are probably the main source from which geneticists and clinicians could benefit. The cases where genomic data are used in healthcare practice vary in complexity and extent of the data used, since the current testing methods are still very expensive and not widely used. We can see simple testing like identifying genes and mutations as well as full sequencing of alleles and the use of micro-arrays to identify the expression of vast number of genes in each individual. Naturally, the Clinical Genomics SIG has been focusing on tests that are routinely done in healthcare, while preparing the information infrastructure standard for more futuristic cases.
>
> The core Genotype model is the GeneticLocus model. It consists of various types of genomic data relating to a specific DNA locus including sequencing, expression and proteomic data. Within the GeneticLocus model HL7 has utilized existing bioinformatics markups to represent raw data received from genomic facilities. Examining and constraining these markups is a work in progress and thus this part of the GeneticLocus model is considered informative as well.
>
> The FamilyHistory model is aimed at describing a patient's pedigree with genomic data. Note that this model utilizes the GeneticLocus model to carry the genomic data for the patient's relatives.

The text adapted from HL7 Version 3.0 Clinical Genomics ends here.

2.1.1 COMPONENT CONSTRAINTS

Table 2-1 Component Constraints

Constraint	Constraint Section
No applicable constraints	

2.1.2 COMPONENT DEPENDENCIES

Table 2-2 Component Dependencies

Standard/HITSP Component	Depends On (Name of standard/HITSP Component that it depends on)	Dependency Type (Pre-condition, Post-condition, General)	Purpose (Reason for this dependency)
HITSP/C90 Family History Decision Support for Genetic Risk Analysis	HITSP/C80 Clinical Document and Message Terminology	General	Consistent use of vocabulary across HITSP specifications
HITSP/C90 Family History Decision Support for Genetic Risk Analysis	HITSP/C83 CDA Content Modules	General	Identifies Content Module Data Elements constrained by this Component to be applied within the exchange

2.2 RULES FOR IMPLEMENTING

2.2.1 DATA MAPPING

This section describes the specific data elements used by this Component. Due to the potentially large number of data elements in a particular standard, only the fields that HITSP is constraining differently from the standard will be described here.

Table 2-3 Data Mapping

Data Element	HITSP/C83 Data Elements	Value Set
FamilyHistory		
subject/patient		
patientPerson	See HITSP/C83 Section 2.2.2.1 Personal Information	
administrativeGenderCode	See HITSP/C83 Section 2.2.2.1 Data Element 1.06 Gender See HITSP/C83 Section 2.2.2.18 Data Element 18.24 Family Member Administrative Gender	See HITSP/C80 Section 2.2.1.2.1.2 Administrative Gender
birthTime	See HITSP/C83 Section 2.2.2.18 Data Element 18.08 Family Member Date of Birth	
raceCode	See HITSP/C83 Section 2.2.2.1 Personal Information Data Element 1.10 Race See HITSP/C83 Section 2.2.2.18 Data Element 18.09 Family Member Race	See HITSP/C80 Section 2.2.1.2.7 Race
ethnicGroupCode	See HITSP/C83 Section 2.2.2.1 Personal Information Data Element 1.11 Ethnicity See HITSP/C83 Section 2.2.2.18 Data Element 18.10 Family Member Ethnicity	See HITSP/C80 Section 2.2.1.2.2 Ethnicity
Relative		
Code	See HITSP/C83 Section 2.2.2.18 Data Eleemnt 18.04 Family Member Relationship	See HITSP/C80 Section 2.2.1.2.5 Family Relation Type
relationshipHolder	see patientPerson above	
subjectOf1	See subjectOf1 on patient below	
subjectOf2	See subjectOf2 on patient below	
subjectOf1		
deceasedEstimatedAge	See HITSP/C83 Section 2.2.2.18 Data Element 18.15 Family Member Age at Death	
livingEstimatedAge	See HITSP/C83 Section 2.2.2.18 Data Element 18.23 Family Member Age	
subjectOf2	See HITSP/C83 Section 2.2.2.18 Data Element 18.11 Family Member Medical History	
clinicalObservation	See HITSP/C83 Section 2.2.2.18 Data Element 18.12 Family Member Condition	
Code	See HITSP/C83 Section 2.2.2.7 Data Element 7.02 Problem Type	See HITSP/C80 Section 2.2.3.1.2 Problem Type
Value	See HITSP/C83 Section 2.2.2.7 Data Element 7.03 Problem Name	See HITSP/C80 Section 2.2.3.1.1 Problem

Data Element	HITSP/C83 Data Elements	Value Set
subject/dataEstimatedAge	See HITSP/C83 Section 2.2.2.18 Data Element 18.13 Family Member Age (at Onset)	
geneticLocus	See HITSP/C83 Section 2.2.2.18 Data Element 18.18 Family Member Genetic Test Information	
Value		See HITSP/C80 Section 2.2.3.13.1 Gene Names
component1/individualAllele		
Value		See HITSP/C80 Section 2.2.3.13.4 Genetic Reference Sequences
component3/sequenceVariation		
Value		See HITSP/C80 Section 2.2.3.13.2 Genetic Sequence Variations
component2/associatedObservation		
Code		Shall contain the code 48002-0 from LOINC. See HITSP/C80 2.2.1.3.11.2 Genetic Test Result Values
Value		Shall contain one of the legal values provided for Genomic Source Class from the table provided in HITSP/C80 2.2.1.3.11.2 Genetic Test Result Values.

2.3 STANDARDS

2.3.1 REGULATORY GUIDANCE

Table 2-4 Regulatory Guidance

Regulation	Description
No applicable regulatory guidance	

2.3.2 SELECTED STANDARDS

Table 2-5 Selected Standards

Standard	Description
Health Level Seven (HL7) Version 3.0 Clinical Genomics; Pedigree, Release 1	The HL7 Clinical Genomic Pedigree is an XML-based message markup standard that specifies the structure and semantics of family history for the purpose of exchange. The Pedigree model is one instantiation of HL7's Version 3.0 Reference Information Model (RIM) into a specific message format. This model further builds upon other HL7 standards beyond just the Version 3.0 Reference Information Model (RIM) and incorporates Version 3.0 Data Structures, Vocabulary, and the XML Implementation Technology Specifications for Data Types and Structures. For more information visit www.hl7.org

2.3.3 INFORMATIVE REFERENCE STANDARDS

Table 2-6 Informative Reference Standards

Standard	Description
Health Level Seven (HL7) Version 2 Implementation Guide: Clinical Genomics; Fully LOINC-Qualified Genetic Variation Model, Release 1	This guide calls for specific vocabulary standards for the exchange of laboratory information. Use of standard vocabularies is important for a number of reasons. Use of standard vocabularies allows broad distribution of healthcare information without the need for individual institutions to exchange master files for data such as test codes, result codes, etc. Each institution maps its own local vocabularies to the standard code, allowing information to be shared broadly, rather than remaining isolated as a single island of information. Standard vocabularies, particularly coded laboratory results, enable more automated decision support for patient healthcare, as well as more automated public health surveillance of populations. For more information visit www.hl7.org

3.0 APPENDIX

The following sections include relevant materials referenced throughout this document.

No additional information at this time.

4.0 CHANGE HISTORY

The following sections provide the history of all changes made to this document since the last publication.

4.1 DECEMBER 10, 2008

The changes in this construct address the following comments received during the Public Comment and Inspection Testing period (September 29 – October 24, 2008).

5451, 5482, 5492

4.1.1 GLOBAL

The following changes were applied through-out the document for consistency with the HITSP suite of Interoperability Specifications.

- The Component name was changed to Family History Decision Support for Genetic Risk Analysis

4.1.2 SECTION 2.2.1 DATA MAPPING

- Updated references to HITSP/C80 sections
- Defined generic variation using HGVS nomenclature instead of Single Nucleotide Polymorphisms
- Modified the last three rows of the table to use correct vocabularies and differentiate between inherited genetic variants tagged with 'germline' and tumor variants (somatic variants).

Minor editorial changes were made to this document.

4.2 DECEMBER 18, 2008

Upon approval by the HITSP Panel on December 18, 2008, this document is now Released for Implementation.

4.3 JUNE 30, 2009

Minor editorial changes were made to this document. Boilerplate text was removed for simplification. The term “actor” was replaced with “interface”.

4.4 JULY 8, 2009

Upon approval by the HITSP Panel on July 8, 2009, this document is now Released for Implementation.