The HL7 Healthcare Connection

An HL7 Overview

HL7 Ambassador Webinar

January 30, 2013

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Intermountain Healthcare Clinical Genetics Institute

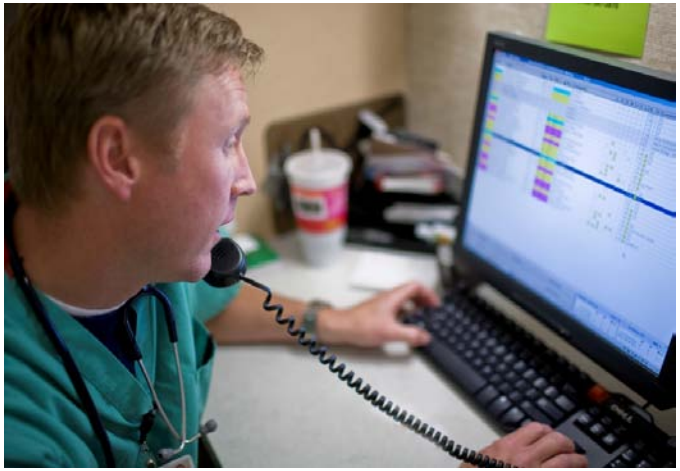
and HL7 Clinical Genomics workgroup

Topics

- Need for Electronic Healthcare Information Exchange
- Healthcare Trends, Challenges, which argue for Standards and their Benefits
- The HL7 Organization
- High-Level Review of Core Standards
- New HL7 Initiatives

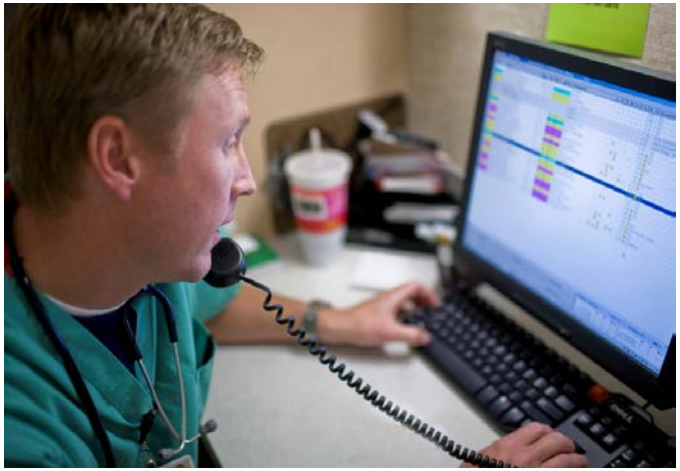
Need For Integrated Systems

Doctors need to be connected with each other – especially during transfer of care



Need For Integrated Systems

Doctors need to be connected with pharmacists – reduce harmful errors



Need For Integrated Systems

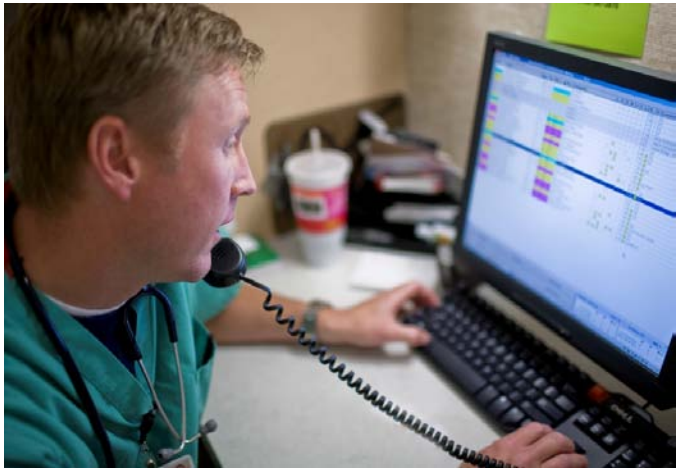
Hospitals need to be connected with each other – especially for medical record transfer

Need For Integrated Systems

Laboratories need to be connected to the patient's electronic health record

Need For Integrated Systems

Doctors need to be connected to the patient's personal health record



Global Healthcare Trends

- Rising cost of healthcare
  - Under or not insured
  - Aging population
  - High cost of chronic care
  - Demand on public health hospitals
  - System and organizational inefficiencies
- Paper to Electronic Records
  - Better clinical outcomes
  - Cost effective
  - Meaningful Use program in the United States
- Public Health
  - Prevention efforts (Immunization, etc.)
  - Bioterrorism and pandemic events (Anthrax, Avian Flu, TB, etc.)

Global Healthcare Trends

- Consumer Empowered
  - Patients and providers seeking greater access and control over information
  - Personal Health Records empower a consumer to manage their own health
- National-Regional IT Networks
  - Canada, Finland, Denmark, Austria, USA, UK, Australia
  - Community Healthcare Information Network
  - Government selected healthcare standards
  - Government-sponsored conformance testing
- Biotech Era
  - Personalized medicine is beginning to emerge, e.g. genomic data and test for cancer drug

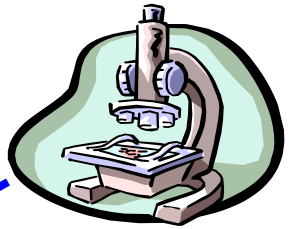
Healthcare Information Exchange Challenges

- *Within* healthcare institutions:
  - How can patient's clinical data from different sources (lab, pharmacy, clinician notes, etc) be brought to patient's point of care and into an electronic medical record?
- *Across* healthcare institutions and others groups needing healthcare data (insurance, public health, research):
  - How can clinical data be shared among different healthcare enterprises using different technology?
  - How can the same patient be identified across different institutions?
  - How can data exchange be secured and access to patient data be monitored?

Many Types of Healthcare Information Need to be Exchanged

Pharmacy Medication Lists

Government Agencies, Public Health, Research



Lab Test Results

Payers / Financial Systems

Patient

Hospitalization Summaries

Home Health Monitoring Devices

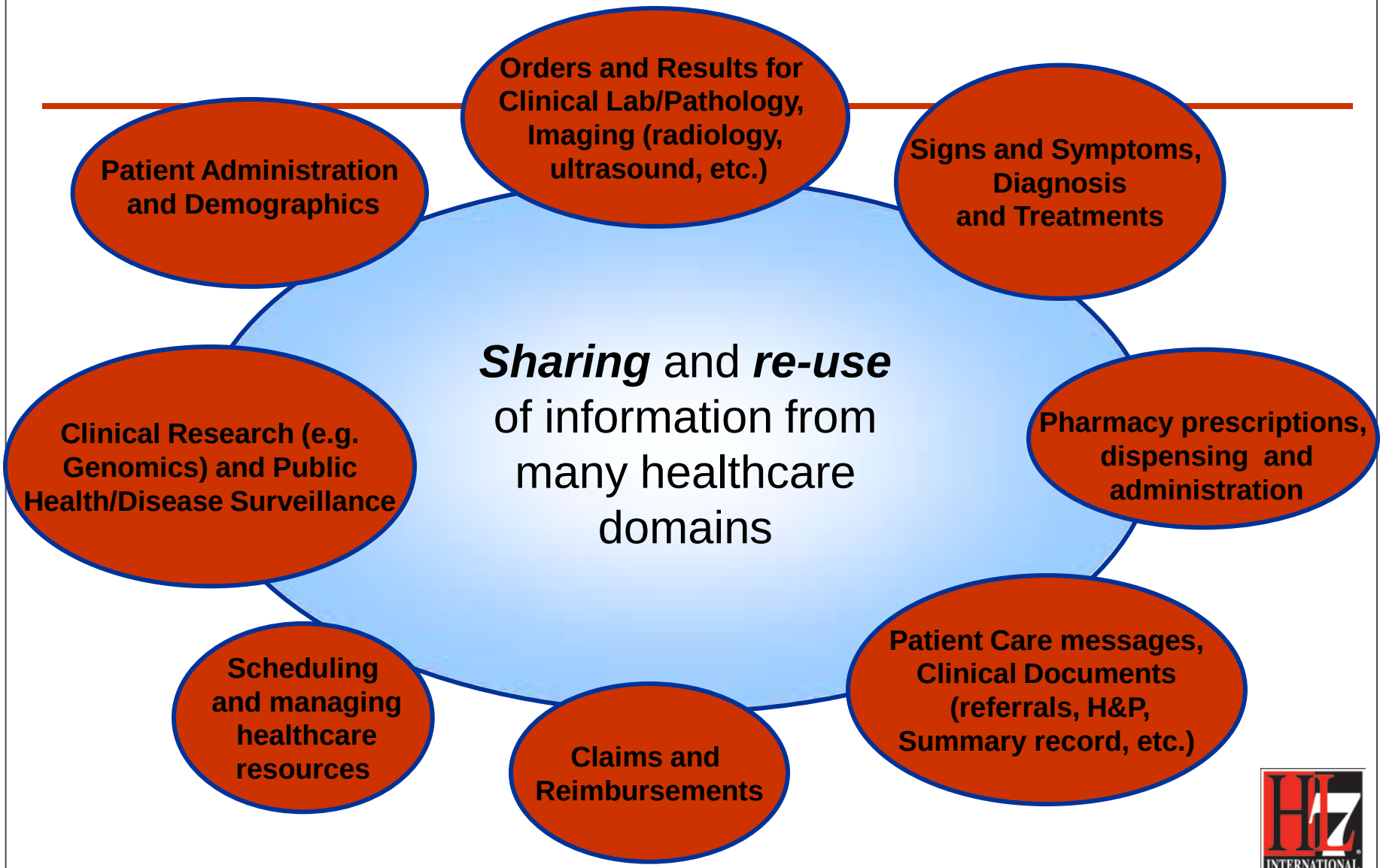
Medical Imaging Results

Doctors Orders and Clinicians Notes

Healthcare IT Stakeholders

- Patients
- Consumers
- General Practitioners
- Specialists
- Outpatient Healthcare Providers
- Residential Care Providers
- Hospitals
- Healthcare Administration
- Pharmaceutical
- Payers, Insurance
- Employers
- Medical Equipment
- Review Boards
- Practice Guidelines
- Government Agencies
- Standards Enforcement Agencies

HL7 Has Produced a Family of Standards



The HL7 Organization

- Founded in 1987, Health Level Seven International (HL7), with members in over 55 countries, is a not-for-profit, ANSI-accredited standards developing organization

- HL7 is 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 management, delivery and evaluation of health services

- HL7's 2,300+ members include approximately 500 corporate members who represent more than 90% of the information systems vendors serving healthcare

- Over 45 healthcare standards from anatomic pathology to vocabulary

Take a Flash tour at

http://www.hl7.org/documentcenter/public/training/IntroToHL7/player.html

HL7 Mission - Interoperability Goals

- HL7's mission is to provide standards for interoperability that:
  - improve care delivery
  - optimize workflow
  - reduce ambiguity
  - enhance knowledge transfer
- Wide range of healthcare standards: clinical, clinical genomics, administrative, clinical research, electronic claims attachments, public health, personal health, etc

HL7 High Level Goals

- Develop coherent, extendible standards that permit structured, coded healthcare information of the type required to support patient care, to be exchanged between computer applications, while preserving the meaning

- Promote the use of HL7 standards worldwide through the creation of HL7 International Affiliate organizations

HL7 High Level Goals

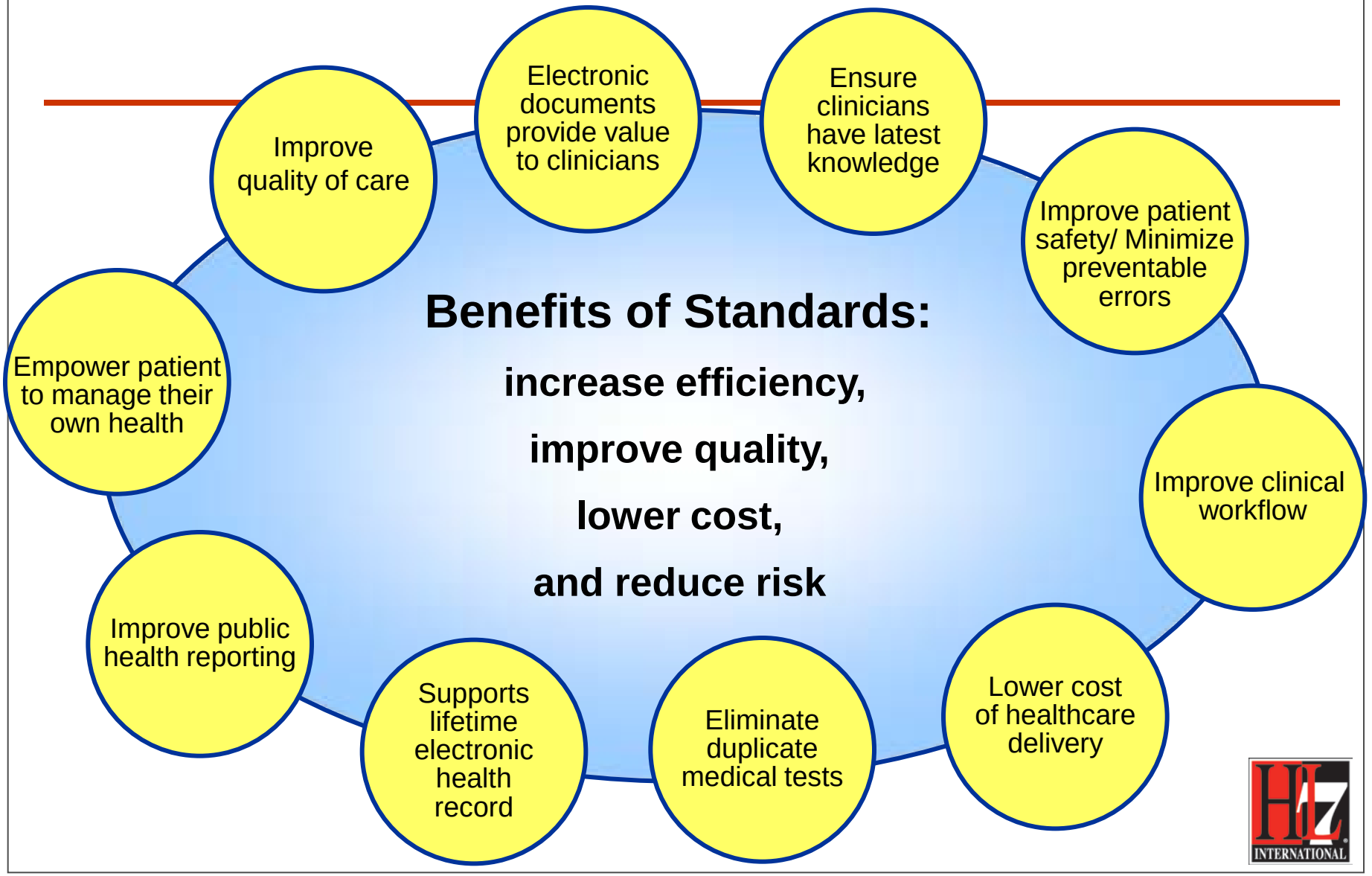
- Stimulate, encourage and facilitate domain experts from healthcare industry stakeholder organizations to participate in HL7 to develop healthcare information standards in their area of expertise

- Collaborate with healthcare information technology users to ensure that HL7 standards meet real-world requirements, and that appropriate standards development efforts are initiated by HL7 to meet emergent requirements

Standards Drive Increased Business for Healthcare IT Vendors and Service Providers

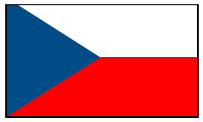
- Speed of development, faster time to market
- Lower development & installation costs, over customized interfaces
- Clients prefer the flexibility of products with standardized interfaces
- Enhanced interoperability of product
- Standards create best practices for the international community
- Bigger market beyond that for proprietary products
- More scalable solution

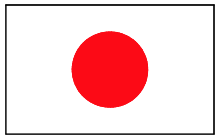
Healthcare Standards Improve Patient Care



An International Organization with Over 30+ HL7 Affiliates


Argentina

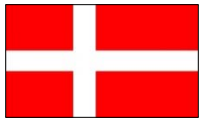

Czech Republic


Japan


New Zealand


Taiwan


Australia

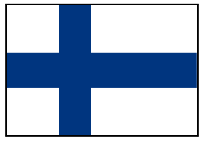

Denmark

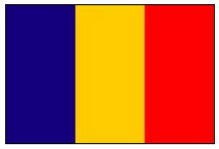

South Korea


The Netherlands


Turkey

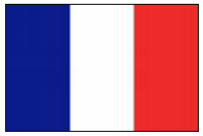

Austria


Finland


Romania


United Kingdom


Brazil


France


Russia

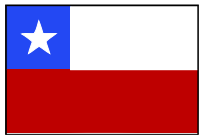

United States


Canada


Germany

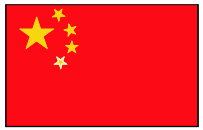

Singapore


Uruguay


Chile


Greece

And growing


China


Hong Kong


Spain


Columbia


India


Luxembourg


Sweden

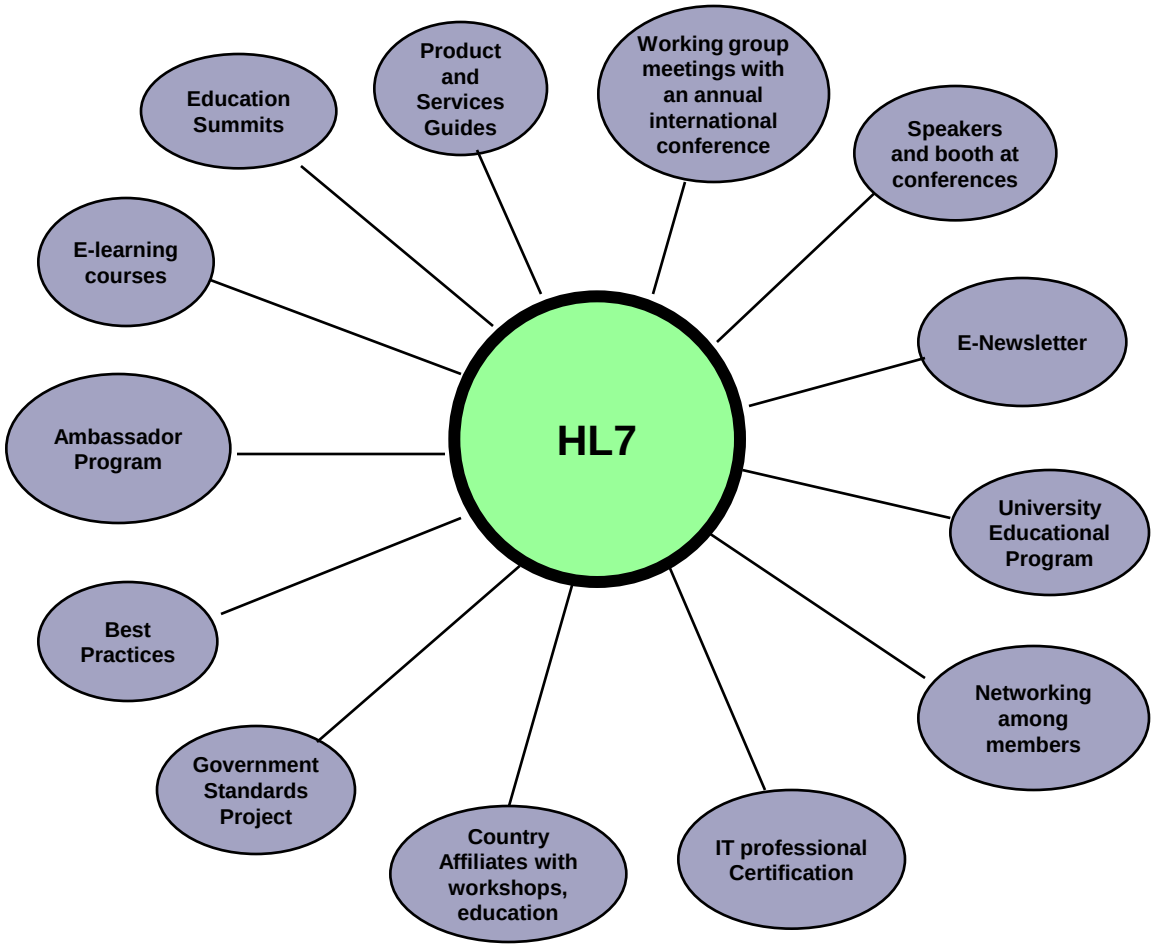

Croatia


Italy


Mexico


Switzerland

Additional HL7 Programs and Activities



Still to Come

- HL7 Family of Standards
  - Version 2 messaging
  - Version 3 messaging
  - The Reference Information Model (RIM)
  - Clinical Document Architecture
  - EHR specifications
  - Clinical Genetics
- Research on annual cost savings when interoperable systems are implemented
- Other products, activities, and benefits HL7 has to offer

HL7 Version 2

DESCRIPTION

- HL7's Version 2.x (V2) messaging standard is the workhorse of electronic data exchange in the clinical domain and arguably the most widely implemented standard for healthcare in the world. This messaging standard allows the exchange of clinical data between systems. It is designed to support a central patient care system as well as a more distributed environment where data resides in departmental systems.

BENEFITS

- Supports the majority of the common interfaces used in the healthcare industry globally
- Provides a framework for negotiations of what is not in the standard
- Reduces implementation costs
- Generally backward compatible
- 95% of US healthcare organizations use HL7 V2.x
- More than 35 countries have HL7 V2.x implementations

V2.x Messaging

- HL7 version 2 defines a series of electronic messages. Since 1987 the standard has been updated regularly, resulting in versions 2.1, 2.2, 2.3, 2.3.1, 2.4, 2.5, 2.5.1, 2.6, and 2.7. The v2.x standards are backward compatible . v2.x messages use one-character delimiters.

- The following is an example of an admission record:

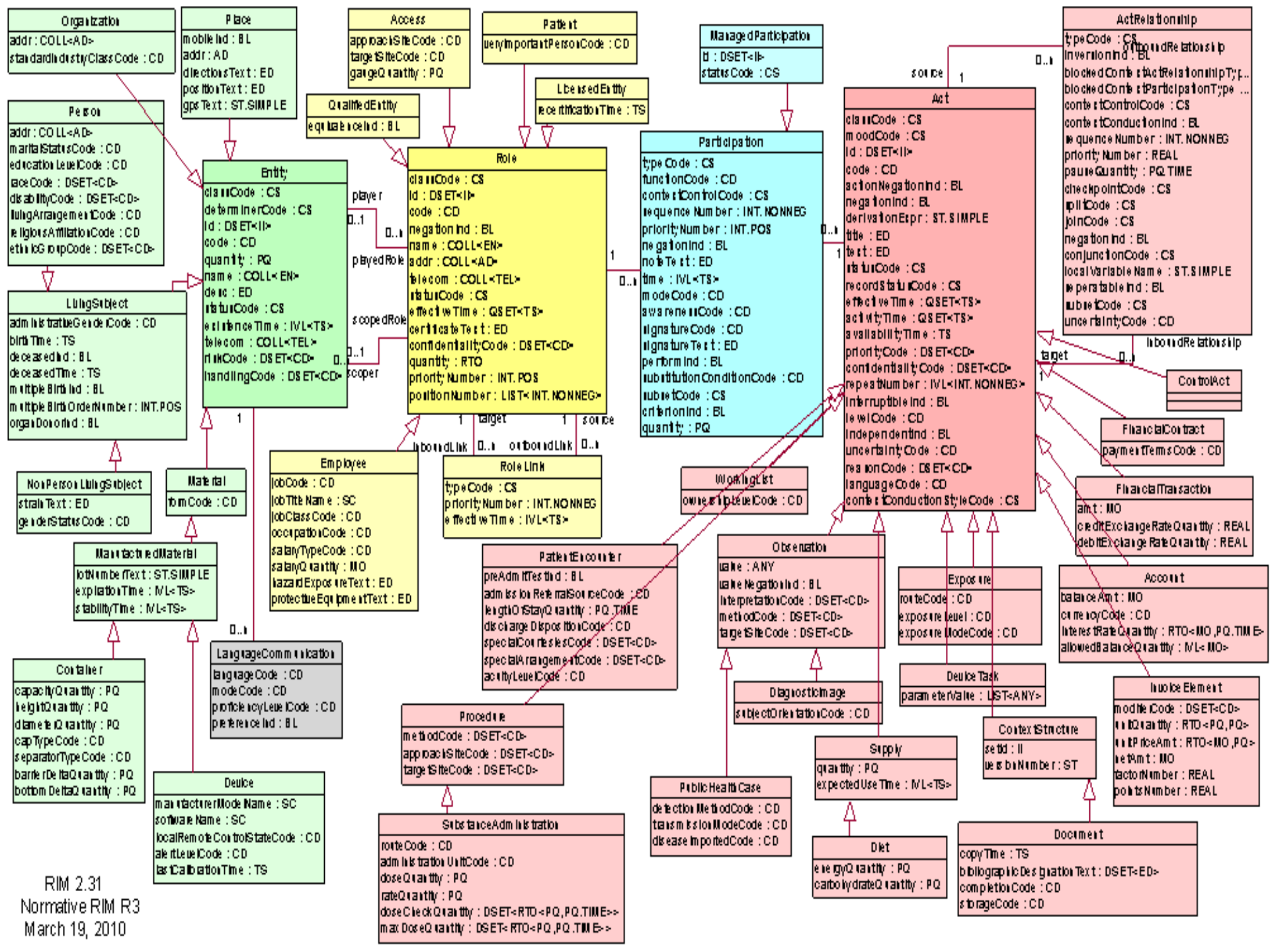
```
MSH|^~\&|MegaReg|XYZHospC|SuperOE|XYZImgCtr|20060529090131-
0500||ADT^A01^ADT_A01|01052901|P|2.5
EVN||200605290901||||200605290900
PID|||56782445^^^UAReg^PI||KLEINSAMPLE^BARRY^Q^JR||19620910|M||2028-
9^^HL70005^RA99113^^XYZ|260 GOODWIN CREST DRIVE^^BIRMINGHAM^AL^35 209^^M~NICKELL'S
PICKLES^10000 W 100TH AVE^BIRMINGHAM^AL^35200^^O ||||||0105I30001^^^99DEF^AN
PV1||I|W^389^1^UABH^^^^3||||12345^MORGAN^REX^J^^^MD^0010^UAMC^L||678
90^GRAINGER^LUCY^X^^^MD^0010^UAMC^L|MED|||||A0||13579^POTTER^SHER
MAN^T^^^MD^0010^UAMC^L|||||||||||||||||||||||||200605290900
OBX|1|NM|^Body Height||1.80|m^Meter^ISO+|||||F
OBX|2|NM|^Body Weight||79|kg^Kilogram^ISO+|||||F
AL1|1||^ASPIRIN
DG1|1||786.50^CHEST PAIN, UNSPECIFIED^I9|||A
```

V3 Messaging

- **The Reference Information Model (RIM)** is the cornerstone of the HL7 Version 3 development process and an essential part of the HL7 V3 development methodology. HL7 v3 messages are based on an XML encoding syntax.
- **Conceptual foundation** – a single, common reference information model to be used across HL7
- **Semantic foundation** – in explicitly defined concept domains drawn from the best terminologies
- **Abstract design methodology** that is technology-neutral – able to be used with whatever is the preferred technology: information resources, documents, messages, services, applications

Five core concepts of the RIM

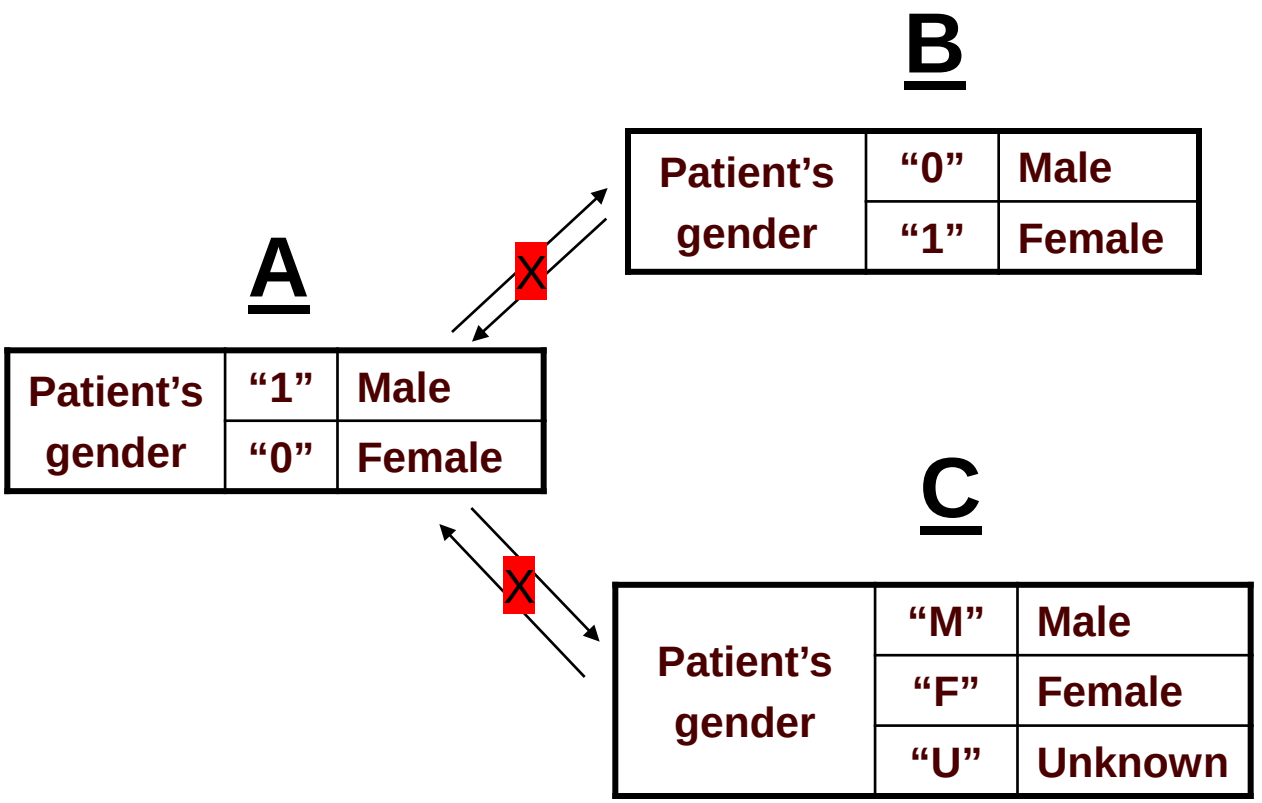
- Every happening is an Act
  - Procedures, observations, medications, supply, registration, etc.
- Acts are related through an ActRelationship
  - composition, preconditions, revisions, support, etc.
- Participation defines the context for an Act
  - author, performer, subject, location, etc.
- The participants are Roles
  - patient, provider, practitioner, specimen, employee etc.
- Roles are played by Entities
  - persons, organizations, material, places, devices, etc.


Organization
addr : COLL<AD>
standardIndustryClassCode : CD
Place
mobileInd : BL
addr : AD
directionsText : ED
positionText : ED
gpsText : ST.SIMPLE
Access
approachSiteCode : CD
targetSiteCode : CD
gaugeQuantity : PQ
Patient
veryImportantPersonCode : CD
ManagedParticipation
id : DSET<II>
statusCode : CS
ActRelationship
typeCode : CS
inversionInd : BL
blockedContextActRelationshipTy...
blockedContextParticipationType ...
contextControlCode : CS
contextConductionInd : BL
sequenceNumber : INT.NONNEG
priorityNumber : REAL
pauseQuantity : PQ.TIME
checkpointCode : CS
splitCode : CS
joinCode : CS
negationInd : BL
conjunctionCode : CS
localVariableName : ST.SIMPLE
repeatableInd : BL
subsetCode : CS
uncertaintyCode : CD
Person
addr : COLL<AD>
maritalStatusCode : CD
educationLevelCode : CD
raceCode : DSET<CD>
disabilityCode : DSET<CD>
livingArrangementCode : CD
religiousAffiliationCode : CD
ethicGroupCode : DSET<CD>
QualifiedEntity
equivalenceInd : BL
LicensedEntity
recertificationTime : TS
Entity
classCode : CS
determinerCode : CS
id : DSET<II>
code : CD
quantity : PQ
name : COLL<EN>
desc : ED
statusCode : CS
existenceTime : IVL<TS>
telecom : COLL<TEL>
riskCode : DSET<CD>
handlingCode : DSET<CD>
player
playedRole
scopedRole
scoper
Role
classCode : CS
id : DSET<II>
code : CD
negationInd : BL
name : COLL<EN>
addr : COLL<AD>
telecom : COLL<TEL>
statusCode : CS
effectiveTime : QSET<TS>
certificateText : ED
confidentialityCode : DSET<CD>
quantity : RTO
priorityNumber : INT.POS
positionNumber : LIST<INT.NONNEG>
Participation
typeCode : CS
functionCode : CD
contextControlCode : CS
sequenceNumber : INT.NONNEG
priorityNumber : INT.POS
negationInd : BL
noteText : ED
time : IVL<TS>
modeCode : CD
awarenessCode : CD
signatureCode : CD
signatureText : ED
performInd : BL
substitutionConditionCode : CD
subsetCode : CS
criterionInd : BL
quantity : PQ
Act
classCode : CS
moodCode : CS
id : DSET<II>
code : CD
actionNegationInd : BL
negationInd : BL
derivationExpr : ST.SIMPLE
title : ED
text : ED
statusCode : CS
recordStatusCode : CS
effectiveTime : QSET<TS>
activityTime : QSET<TS>
availabilityTime : TS
priorityCode : DSET<CD>
confidentialityCode : DSET<CD>
repeatNumber : IVL<INT.NONNEG>
interruptibleInd : BL
levelCode : CD
independentInd : BL
uncertaintyCode : CD
reasonCode : DSET<CD>
languageCode : CD
contextConductionStyleCode : CS
source
target
inboundRelationship
outboundRelationship
ControlAct
FinancialContract
paymentTermsCode : CD
FinancialTransaction
amt : MO
creditExchangeRateQuantity : REAL
debitExchangeRateQuantity : REAL
Account
balanceAmt : MO
currencyCode : CD
interestRateQuantity : RTO<MO,PQ.TIME>
allowedBalanceQuantity : IVL<MO>
InvoiceElement
modifierCode : DSET<CD>
unitQuantity : RTO<PQ,PQ>
unitPriceAmt : RTO<MO,PQ>
netAmt : MO
factorNumber : REAL
pointsNumber : REAL
LivingSubject
administrativeGenderCode : CD
birthTime : TS
deceasedInd : BL
deceasedTime : TS
multipleBirthInd : BL
multipleBirthOrderNumber : INT.POS
organDonorInd : BL
NonPersonLivingSubject
strainText : ED
genderStatusCode : CD
Material
formCode : CD
ManufacturedMaterial
lotNumberText : ST.SIMPLE
expirationTime : IVL<TS>
stabilityTime : IVL<TS>
Container
capacityQuantity : PQ
heightQuantity : PQ
diameterQuantity : PQ
capTypeCode : CD
separatorTypeCode : CD
barrierDeltaQuantity : PQ
bottomDeltaQuantity : PQ
Device
manufacturerModelName : SC
softwareName : SC
localRemoteControlStateCode : CD
alertLevelCode : CD
lastCalibrationTime : TS
LanguageCommunication
languageCode : CD
modeCode : CD
proficiencyLevelCode : CD
preferenceInd : BL
Employee
jobCode : CD
jobTitleName : SC
jobClassCode : CD
occupationCode : CD
salaryTypeCode : CD
salaryQuantity : MO
hazardExposureText : ED
protectiveEquipmentText : ED
RoleLink
typeCode : CS
priorityNumber : INT.NONNEG
effectiveTime : IVL<TS>
inboundLink
outboundLink
WorkingList
ownershipLevelCode : CD
PatientEncounter
preAdmitTestInd : BL
admissionReferralSourceCode : CD
lengthOfStayQuantity : PQ.TIME
dischargeDispositionCode : CD
specialCourtesiesCode : DSET<CD>
specialArrangementCode : DSET<CD>
acuityLevelCode : CD
Procedure
methodCode : DSET<CD>
approachSiteCode : DSET<CD>
targetSiteCode : DSET<CD>
SubstanceAdministration
routeCode : CD
administrationUnitCode : CD
doseQuantity : PQ
rateQuantity : PQ
doseCheckQuantity : DSET<RTO<PQ,PQ.TIME>>
maxDoseQuantity : DSET<RTO<PQ,PQ.TIME>>
Observation
value : ANY
valueNegationInd : BL
interpretationCode : DSET<CD>
methodCode : DSET<CD>
targetSiteCode : DSET<CD>
DiagnosticImage
subjectOrientationCode : CD
PublicHealthCase
detectionMethodCode : CD
transmissionModeCode : CD
diseaseImportedCode : CD
Exposure
routeCode : CD
exposureLevel : CD
exposureModeCode : CD
DeviceTask
parameterValue : LIST<ANY>
Supply
quantity : PQ
expectedUseTime : IVL<TS>
Diet
energyQuantity : PQ
carbohydrateQuantity : PQ
ContextStructure
setId : II
versionNumber : ST
Document
copyTime : TS
bibliographicDesignationText : DSET<ED>
completionCode : CD
storageCode : CD
RIM 2.31
Normative RIM R3
March 19, 2010

Domains in the Normative HL7 V3 standard

- Accounting & Billing
- Claims & Reimbursement
- Materials Management
- Patient Administration
- Personnel Management
- Scheduling
- Blood bank
- Care Provision
- Clinical Decision Support
- Clinical Document Architecture
- Clinical Genomics
- Diagnostic Imaging
- Immunization
- Laboratory
- Medical Records
- Medication
- Orders and Observation
- Pharmacy
- Public Health
- Regulated Products
- Regulated Studies
- Specimen
- Therapeutic Devices

Need a Standard Coding, Terminology, and Vocabulary System for Common Understanding



A and **B** differ syntactically and cannot interoperate without translation.

A and **C** differ semantically

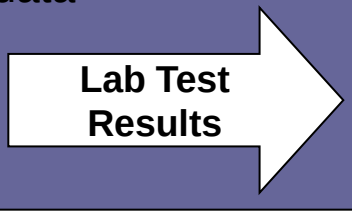
A cannot represent the concept "Unknown"

HL7 Messages and Documents

Messages

- A message is event driven and includes a specific workflow.

- It could include bi-directional flow of data



Documents

- The Clinical Document Architecture (CDA) can facilitate clinical document exchange within and between medical institutions.
- CDA can be used to a bring patient's clinical documents into to patient-centric EHR.
- A collection of information about an encounter
- Can be digitally signed

Clinical Document Architecture (CDA)

- Interoperability
  - An approved standard way to exchange dictated, scanned, or electronic reports on a patient between various health information technology systems and platforms
  - Human readable
    - The “paper world” of clinical documents, forms, etc.
  - Computer readable
    - XML representation of document data
    - EHR discrete data storage
    - Clinical decision support

CDA is the Basis For …

- Consult Note
- Continuity of Care Document
- Diagnostic Imaging Report
- Discharge Summary
- Healthcare-associated Infections, Public Health Case Reports
- History and Physical
- Operative Note
- Personal Health Monitoring
- Plan-2-Plan Personal Health Record
- Quality Reporting Document
- Unstructured Documents
- Emergency Care Summary
- Summary Documents Using HL7 CCD
- Patient Level Quality Data Document Using IHE Medical Summary (XDS-MS)
- Encounter Document constructs
- Consult and History & Physical Note Document
- Immunization Document
- Scanned document
- … and many more …

What is a Continuity of Care Document?

- A medical summary representing the continuity of care record core data set covering one or more healthcare encounters.
- A snapshot in time for a patient, in CDA form, containing the pertinent:
  - clinical,
  - demographic, and
  - administrative data

CCD Required Sections

- Conditions (Problems)
  - active
  - resolved
  - chief complaint
  - reason for visit
  - diagnoses
    - admission
    - discharge
    - pre-operative
    - post-operative
- Allergies and Intolerances
  - pharmacy
  - dietary
  - general
- Medications
  - history
  - administered
  - discharge
  - current

CCD Optional Sections

- Advanced Directives
- Functional Status
- Procedures
- Encounters
- Family History
- Social History
- Immunizations
- Vital Signs
- Fetal Vital Signs
- Lab Results
- Plan of Care

ARUP Laboratories
500 Chipeta Way - Salt Lake City, UT 84108
(800) 522-2787 - www.aruplab.com
Sherrie L. Perkins, MD, Laboratory Director

*** Example Report ***

DOB/Sex: -58 Female
Printed: 07-Oct-10 09:35:48

Procedure	Result	Units	Ref Interval	Accession	Collected	Verified
HFE PCR Specimen	Whole Blood				25-Sep-10 10:15:00	01-Oct-10 13:03:26
C282Y Hemochromatosis Mutation	Negative				25-Sep-10 10:15:00	01-Oct-10 13:03:26
H63D Hemochromatosis Mutation	Negative				25-Sep-10 10:15:00	01-Oct-10 13:03:26
S65C Hemochromatosis Mutation	Negative				25-Sep-10 10:15:00	01-Oct-10 13:03:26
Hemochromatosis Mutation Interpretation	See Note f				25-Sep-10 10:15:00	01-Oct-10 13:03:26

25-Sep-10 10:15:00 Hemochromatosis Mutation Interpretation
Hemochromatosis Interpretive Results:
Negative WT:

C282Y: Negative -- The patient is negative for the HFE C282Y mutation.

H63D: Negative -- The patient is negative for the HFE H63D mutation.

S65C: Negative -- The patient is negative for the HFE S65C mutation.

Mutations in unidentified genes or other mutations in the HFE gene are not ruled out.

25-Sep-10 10:15:00 HFE PCR:
Client Accession number:
Is the Patient Fasting? YES
25-Sep-10 10:15:00 Hemochromatosis Mutation Interpretation:

This result has been reviewed and approved by Hunter Best, Ph.D.

25-Sep-10 10:15:00 Hemochromatosis Mutation Interpretation:
BACKGROUND INFORMATION: Hemochromatosis (HFE) 3 Mutations

CHARACTERISTICS: Disorder of iron metabolism resulting in excessive iron storage leading to increased skin pigmentation, arthritis, hypogonadism, diabetes mellitus, heart arrhythmias/failure, cirrhosis and liver carcinoma.
INCIDENCE: One in 300 individuals of Northern European descent; unknown in other ethnicities.
INHERITANCE: Autosomal recessive.
PENETRANCE: 5 percent ofC282Y homozygotes, 1 percent of C282Y/H63D compound heterozygotes and rare H63D homozygotes develop clinical symptoms.
CAUSE: Two pathogenic HFE gene mutations on opposite chromosomes.
MUTATIONS TESTED: p.C282Y (c.845G>A), p.H63D (c.187C>G), and p.S65C (c.193A>T).
CLINICAL SENSITIVITY: 85 percent of hereditary hemochromatosis in Northern Europeans is caused by C282Y homozygosity and 5 percent by C282Y/H63D compound heterozygosity.
METHODOLOGY: PCR and fluorescence monitoring.
ANALYTICAL SENSITIVTY AND SPECIFICITY: 99 percent.
LIMITATIONS: HFE mutations, other than those targeted, will not be detected. Rare diagnostic errors may occur due to primer site mutations.
This test is performed pursuant to an agreement with BioRad Laboratories, Inc.

The performance characteristics of this test were validated by ARUP Laboratories. The U.S. Food and Drug Administration (FDA) has not approved or cleared this test. However, FDA approval or clearance is currently not required for clinical use of this test. The results are not intended to be used as the sole means for clinical diagnosis or patient

Genetic Testing Report

[ClinicalDocument: templateId 2.16.840.1.113883.10.20.20]

The GeneticTestingReport is a document template and thus serves as the root template for the GTR Implementation Guide. Its organization is described in the Approach section of this document. The sub-sections residing here constitute the backbone of the GTR. A specific genetic test is described in the TestDetailsSection which serves as a blueprint specialized sections describing testing like genetic variation or gene expression.

1. **SHALL** contain exactly one [1..1] `code/@code="51969-4"` *Genetic analysis summary report* (CodeSystem: 2.16.840.1.113883.6.1 LOINC) (CONF-GTR-1)
2. **SHALL** contain exactly one [1..1] `title` (CONF-GTR-7)
   - Default title is "Genetic Testing Report".
3. **SHALL** contain exactly one [1..1] `component`
   a. Contains exactly one [1..1] *Summary Section* (templateId: 2.16.840.1.113883.10.20.20.1.1)
4. Contains at least one [1..*] `component`
   a. Contains exactly one [1..1] *Test Details Section* (templateId: 2.16.840.1.113883.10.20.20.1.8)
5. Contains zero or one [0..1] `component`
   a. Contains exactly one [1..1] *Test Information Section* (templateId: 2.16.840.1.113883.10.20.20.1.9)
6. Sections and subsections **SHALL** have a title and the title **SHALL NOT** be empty. Text of a section title can specialize the section code by being more specific, for example, a hearing loss genetic testing report.
7. Sections **SHALL** appear in the order they are presented in this guide. Thus, SummarySection which **SHALL** appear first and TestInformationSection which **SHOULD** appear last. In between, TestDetailsSection can be repeated per the no. of genetic tests performed. Note that a TestInformationSection can appear in each of the specific test sections.

```
<?xml version="1.0" encoding="UTF-8"?>
<ClinicalDocument xmlns:xsi="http://www.w3.org/2001/XMLSchema-instance"
 xmlns="urn:hl7-org:v3" xsi:schemaLocation="urn:hl7-org:v3 CDA.xsd">
  <typeId root="2.16.840.1.113883.1.3" extension="POCD_HD000040"/>
  <templateId root="2.16.840.1.113883.10.20.20"/>
  <id root="2.16.840.1.113883.18.12.7.30.9.1" extension="c266"/>
  <code code="51969-4" codeSystem="2.16.840.1.113883.6.1"
 codeSystemName="LOINC" displayName="Genetic analysis summary report"/>
  <title>Genetic Testing Report</title>
  <effectiveTime value="20100809"/>
```

Dilated Cardiomyopathy Panel B - 5 Gene Panel Test Report

Patient	Patrick Pump		
Date of birth	May 5, 1947	**Sex**	Male
Contact info	address not available Telecom information not available	**Patient IDs**	123456789 2.16.840.1.113883.18.12.7.30.9.2
Document Id	c266 2.16.840.1.113883.18.12.7.30.9.1		
Document Created:	August 9, 2010		
Author	Jean Genome,		
Legal authenticator	Jean Genome of HPCGG Laboratory for Molecular Medicine signed at February 12, 2006		
Document maintained by	2.16.840.1.113883.19.3.2409		

Table of Contents

Summary Section

Indications

- Clinical Diagnosis and Family History of DCM

Specimen and Genomic Source

- Peripheral Blood
- Genomic source class: Germline

Tests Performed

- Dilated Cardiomyopathy Panel B (5 genes)

Overall Interpretation

- **Positive.** DNA sequencing of the coding regions and splice sites of the ACTC, LDB3, LMNA, PLN and TAZ genes revealed a heterozygous R377C variant in exon 6 of the LMNA gene (NM_170707.1). The R377C variant has been reported in the literature (Muchir 2000, Ki 2002, Kubben 2006, van Tintelen 2007). As such, this variant is highly likely to be pathogenic and therefore causative for DCM. Genetic testing of this patient's biological parents and other family members, particularly those who are affected, may help to confirm the significance of this variant. Please note that the laboratory can attempt testing on tissue specimens from deceased family members. It should be noted that the expression of DCM is the product not only of a DCM gene variant, but also of other modifier genes and environmental factors. The significance of a variant should always be interpreted in the context of the patient's clinical manifestations. COMMENTS: Common sequence variants of unlikely clinical significance are not included in this report but are available upon request.

Recommendations

- If you would like more information about the clinical manifestations of DCM variants we recommend you visit a cardiology center with expertise in the management of dilated cardiomyopathy such as the BWH Cardiovascular Genetics Center at 617-732-4837 (www.brighamandwomens.org/cvcenter/Services/genetics.asp). DCM caused by LMNA variants is inherited in an autosomal dominant manner where each first-degree relative of an individual with a DCM causing mutation has a 50% (or 1 in 2) chance of inheriting the mutation. Genetic testing is available for at-risk family members if desired. Genetic counseling is recommended for this patient and his family. For assistance in locating nearby genetic counseling services please call the laboratory at 617-768-8500 or email at LMM@partners.org.

Test Information

Background

- Dilated cardiomyopathy (DCM) is characterized by ventricular chamber enlargement and systolic dysfunction with normal left ventricular wall thickness. The estimated prevalence of DCM is 1/2,500 and about 20-35% of cases have a family history showing a predominantly autosomal mode of inheritance. Mutations in more than 20 genes have been shown to cause DCM, several of which (including MYH7, MYBPC3, TNNT2, TNNI3, TPM1 and ACTC), are also known to cause hypertrophic cardiomyopathy. Mutations in some genes cause additional abnormalities: Lamin A/C (LMNA) mutations are frequently found in DCM that occurs with progressive conduction system disease. Mutations in the Tafazzin (TAZ) gene cause Barth syndrome, an X-linked cardioskeletal myopathy in infants. In addition, mutations in several genes (including LMNA, DES, SGCD and EMD) can cause DCM in conjunction with skeletal myopathy. Genetic testing can confirm the diagnosis of DCM in patients with disease as well as identify at risk family members prior to the onset of symptoms.

The EHR-S Functional Model

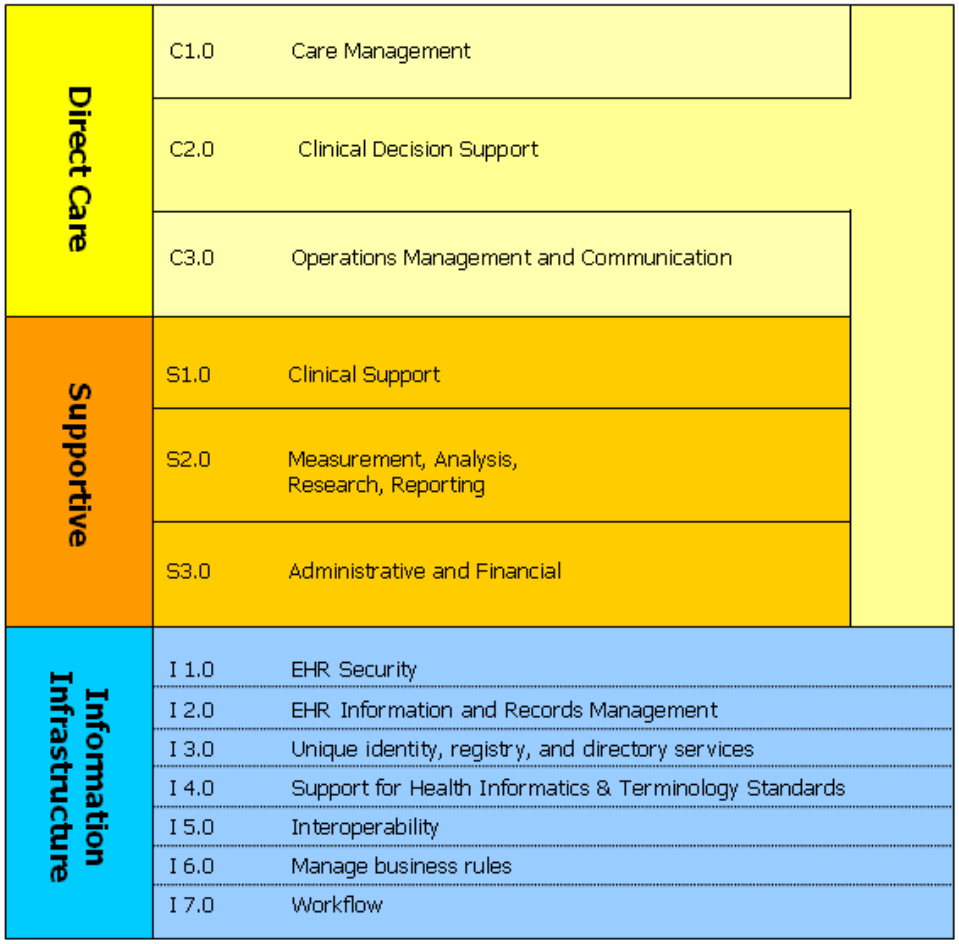
Is…

- A system specification
- An EHR system specification
- A reference list of functions that may be present in an EHR-S (the "what")
  - Enables consistent expression of functionality
  - Provides flexibility for innovation and product differentiation
  - Gold standard, sensitive to what can practically be done by a system, future system development

Is Not…

- A messaging specification
- An EHR specification
- An implementation specification (not the "how")
  - Does not prescribe technology
  - Does not dictate how functions must be implemented (e.g., via the user interface, database design)

EHR-S Functional Model at a Glance



Functions describe the behavior of a system in user-oriented language so as to be recognizable to the key stakeholders of an EHR System

EHR-S Profiles Developed or Under Development

- Emergency Department
- Child Health
- Long Term Care
- Behavioral Health
- Records Management & Evidentiary Support
- Regulated Clinical Research (Clinical Trials)
- Vital Statistics Reporting

For more information:

HL7 Electronic Health Record
http://www.hl7.org/ehr/index.asp

HL7 Functional Profile Registry
http://xreg2.nist.gov:8080/ehrsRegistry/index.jsp

Interoperability Between Hospital-Based Outpatient Clinicians and External Laboratories

Annual savings of $31.8 billion at highest level of interoperability. In addition to reducing duplicate tests, it would –

1) reduce delays and costs associated with paper-based ordering and reporting of results,
2) provider-laboratory connectivity would give clinicians better access to patients' longitudinal test results,
3) eliminate errors associated with reporting results orally,
4) optimize ordering patterns by making information on test costs readily available to clinicians, and
5) make testing more convenient for patients.

Walker, et al. The Value Of Health Care Information Exchange And Interoperability *Health Affairs* Web Exclusive, January 19, 2005

Connectivity Between Office-Based Clinicians and External Radiology Centers

Annual savings of $26.2 billion at highest level of interoperability. In addition to reducing duplicate tests, it would –

1) save time and costs associated with paper- and film-based processes,
2) improve ordering by giving radiologists access to relevant clinical information, thereby enabling them to recommend optimal testing,
3) improve patient safety by alerting both the provider and the radiologist to test contraindications,
4) facilitate coordination of care and help prevent errors of omission by enabling automated reminders when follow-up studies are indicated, and
5) lessen adverse environmental impacts by reducing the use of chemicals and paper in film processing.

Walker, et al. The Value Of Health Care Information Exchange And Interoperability *Health Affairs* Web Exclusive, January 19, 2005

Interoperability Between Outpatient Providers and Pharmacies

Annual savings of $ 2.71 billion at highest level of interoperability. In addition to reducing the number of medication-related phone calls for both clinicians and pharmacists, it would –

1) improve clinical care by facilitating the formation of complete medication lists, thereby reducing duplicate therapy, drug interactions and other adverse drug events, and medication abuse,
2) enable automated refill alerts,
3) offer clinicians easy access to information about whether patients fill prescriptions,
4) complete insurance forms required for some medications,
5) help identify affected patients in the event of drug recalls, uncover new side effects, and improve formulary management.

Walker, et al. The Value Of Health Care Information Exchange And Interoperability
Health Affairs Web Exclusive, January 19, 2005

Provider to Provider Connectivity

Annual savings of $13.2 billion at highest level of interoperability. In addition to saving time associated with handling chart requests and referrals it would –

1) would reduce fragmentation of care from scattered records and improve referral processes.

Walker, et al. The Value Of Health Care Information Exchange And Interoperability
Health Affairs Web Exclusive, January 19, 2005

Use Case Medium-Size Hospital

The hospital (with 50–199 beds) would invest $2.7 million in clinical systems and interfaces to achieve the highest level of interoperability. After the first year, spending $250,000 per year to maintain those systems it would accrue benefits of $1.3 million annually, from

1) its transactions with other providers ($570,000),
2) laboratories ($200,000),
3) radiology centers ($170,000),
4) payers ($250,000), and
5) pharmacies ($70,000).

Walker, et al. The Value Of Health Care Information Exchange And Interoperability
Health Affairs Web Exclusive, January 19, 2005

Summary

- Need for computable and interoperable healthcare information
- Standards are critical for exchanging electronic healthcare information
- HL7 is the key organization for producing relevant global healthcare information standards

Working Group Meetings

Health Level Seven® International

May 2013 Working Group Meeting

Atlanta, GA

May 5-10, 2013

[HL7.org]

Home

Cover Page

Hotel Information

Sheraton Atlanta Hotel

Reserve Your Room Today!

Reserve Your Government Room Today!

Meeting Information

Key Information

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Additional Information and Registration Coming!

Ambassador Webinars

Health Level Seven® International

The HL7 Healthcare Connection - An HL7 Overview

Online Webinar

January 30, 2013 1 - 2 pm EST

The HL7 Healthcare Connection - An HL7 Overview

Join us for a Webinar on January 30

REGISTER NOW

Space is limited.
Reserve your Webinar seat now at:
https://www2.gotomeeting.com/register/505905066

Grant Wood, senior IT strategist with Intermountain Healthcare's Clinical Genomics Institute, HL7 ambassador and member of the HL7 Clinical Genomics Work Group, will discuss how the implementation of HL7 standards and messaging architecture solves the problems of disconnected healthcare systems and serves as a vehicle for interoperability with disparate healthcare IT systems, applications and data architectures.

HL7's healthcare standards play a key role in the exchange of electronic data in much of today's global healthcare community and represent some of the most widely implemented healthcare standards in the world. This key role has been expanded in the United States, as many HL7 data models and messaging standards have been chosen to be the foundation of several Meaningful Use Stage 2 requirements. HL7 standards provide a comprehensive framework that improves healthcare delivery, optimizes both clinical and administrative workflow, creates a shared language, and enhances knowledge transfer among all healthcare stakeholders, including healthcare providers and their patients, government agencies, the vendor community, and other related standards groups.

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ACTIVE WORK GROUPS

A
- Affiliate Due Diligence
- Anatomic Pathology
- Architectural Review
- Arden Syntax
- Attachments

C
- Child Health
- Clinical Context Object Workgroup
- Clinical Decision Support
- Clinical Genomics
- Clinical Interoperability Council
- Clinical Statement
- Community Based Collaborative Care

D
- Domain Experts Steering Division

E
- Education
- Electronic Health Records
- Electronic Services
- Emergency Care

F
- Financial Management
- Foundation and Technology Steering Division

G
- Generation of Anesthesia Standards
- Governance and Operations
- Government Projects

H
- Health Care Devices

I
- Imaging Integration
- Implementable Technology Specifications
- Implementation / Conformance
- Infrastructure and Messaging
- International Council
- International Mentoring Committee

PROJECTS

Bridging the Chasm and the CIIC

Terminfo Project

Transition Planning

NLM Contract

WORK GROUP RESOURCES

Decision Making Practices

Project Scope Statement and Project Approval Process

Word template for the Project Scope statement and HL7's Project Approval Process.

Request Rational Software Modeler

Health Level Seven International

Subscribe to List Services

Not logged in

Welcome

You must supply all required fields and can choose as many list services to subscribe to as you would like. At the bottom of this form, you must confirm your agreement to the regulations governing the proper use of our list services and submit this information. You will later receive an email that will provide a link to a page for final confirmation of your desire to subscribe to these lists.

After filling in the basic information below and choosing the lists to which you wish to subscribe, scroll down to the bottom of this page to affirm you understand the proper use of our lists and click the request subscriptions button.

First Name: *

Last Name: *

Organization: *

EMail: *

List Service Password: * (Minimum of 5 characters, maximum of 20)

A password is required when signing up for any lists as a security measurement. This "List service password" may be the same as or different from your HL7 membership password. Make sure you keep this password in a safe place for later login access to the List Manager website. To learn more about passwords, click here for FAQ information

Type of Delivery:

- ○ Mail
- ○ Digest *
- ○ Digest with Attachments

If you are not sure on which type of mail delivery you would like, look at the FAQ's in the Welcome menu for detailed information or click here to jump to that information. **All the lists you choose on this form will all be of this type.**

List Selection:

Anatomic Pathology

- ☐ anatomicpath — Primary List

Architectural Review

- ☐ arb — Primary List

Arden Syntax

- ☐ ardensyntax — Primary List

Attachments

- ☐ asig — Primary List
- ☐ hl7x12ddcp — X12 Claim/Attachment Data Determination Coordination Project

Child Health

- ☐ childhealth — Primary List

wiki.hl7.org

page | discussion | view source | history

Main Page

This is the Main Page of the **HL7 Wiki**, hosted by HL7 Inc, whose main web page is at http://www.hl7.org. The HL7 Wiki is a collaborative technology used to support the HL7 organization. The contents of this Wiki are non binding, see the HL7 website for persistent documents (minutes, standards, etc.). The Wiki currently has 5,705 pages with substantive content. See Basic Editing and Help Resources for information about editing the Wiki, subject to the HL7 Wiki Acceptable Use Policy. To access information, either use the search option or the main page of a HL7 work group or project as shown below.

WGs/Other Groups

- Infrastructure Topic Groups
  - Conformance and Guidance for Implementation/Testing WG
  - Infrastructure and Messaging WG
  - ITS WG
  - Mobile Health WG
  - Modeling and Methodology WG
  - RIMBAA WG (v3 implementers)
  - Security WG
  - Service Oriented Architecture WG
  - Structured Documents WG
  - Templates WG
  - Vocabulary WG
- Health & Clinical Topic Groups
  - Anatomic Pathology WG
  - Arden Syntax WG
  - Attachments WG
  - Child Health WG
  - Clinical Decision Support WG
  - Clinical Genomics WG
  - Clinical Interoperability Council (CIC)
  - Clinical Quality Information WG
  - Clinical Statement WG
  - Community-Based Collaborative Care (CBCC) WG
  - Electronic Health Record (EHR) WG
    - EHR Interoperability (Sub-WG)
    - EHR Records Management & Evidentiary Support (RM-ES) (Sub-WG)
  - Emergency Care WG
  - Generation of Anesthesia Standards WG
  - Government Projects WG
  - Health Care Devices WG
  - Imaging Integration WG

WGs/Other Groups

- Health & Clinical Topic Groups (cont.)
  - Orders & Observations WG
  - Patient Care WG
  - Patient Safety WG
  - Pharmacy WG
  - Public Health and Emergency Response WG
  - Regulated Clinical Research Info. Mgmt. WG (RCRIM)
- Administrative Topic Groups
  - Financial Management WG
  - Patient Administration WG
- Functional Support Groups
  - Education Committee
  - Electronic Services
  - Process Improvement Committee (PIC)
  - Project Services Work Group
  - V2.x Publishing Work Group
  - V3 Publishing Work Group
  - Tooling Work Group
- HL7 Governance Groups
  - Architecture Board
  - Board of Directors of HL7 International
  - International Council
  - Marketing Committee
  - Organizational Relations Committee
  - Policy Advisory Committee
  - Roadmap Committee
  - Technical Steering Committee
    - Domain Experts SD
    - Foundation and Technology SD
    - Structure & Semantic Design SD
    - Technical and Support Services SD

FAQs and Related

- WGMs
  - **Upcoming WGM information and *detailed agendas***
  - Co-Chair Election Schedule
  - Unofficial HL7 Significant others & spouses Auxiliary
- FAQs
  - Best Practices
  - Style Guides & Templates
  - "How to ..." pages
  - V3 Documents (CDA) - FAQ
  - V3 Tooling - FAQ
  - V2 Messaging - FAQ
  - V3 Messaging - Implementation FAQ
  - HL7 OID Registry Frequently Asked Questions
- See also
  - Find or Be a Volunteer for an HL7 Work Group
  - WG Information (HL7.org website)
  - PBS Metrics (Projects, Ballots, and Standards) on GForge (no login required) at TSC Projects > Files > PBS Metrics
  - Search for HL7 Projects
- Related Wikis
  - HL7 TSC Wiki
  - HDF development wiki
  - IHE Wiki
  - Biomed GT Collaborative Terminology Development Wiki
  - MITA project wiki
  - Wikis: HL7 AU, HL7 DE

HL7 Overview

Consolidated CDA

- The development of a single implementation guide that represents harmonization of Health Story guides, HITSP C32, part of the IHE Patient Care Coordination, and the original CCD by HL7
- 9 different types of commonly used CDA documents
  - Continuity of Care Document
  - Consultation Notes
  - Discharge Summary
  - Imaging Integration, and DICOM Diagnostic Imaging Reports
  - History and Physical
  - Operative Note
  - Progress Note
  - Procedure Note
  - Unstructured Documents

Fast **H**ealth **I**nteroperability **R**esources

- V3 puts needs of the modeler before the needs of the implementer
  - New methodology
  - New tools
  - New publishing approach
  - Still built on the RIM, vocabulary & data types, but more hidden

FHIR Resources

- Administrative Concepts
  - Person, Patient, Organization, Device, Facility
  - Coverage, Invoice, etc.
- Clinical Concepts
  - Allergy, Problem, Medication, Family History
  - Care Plan
- Infrastructure things
  - Document, Message, Conformance/Profiling

Mobile Health

- If someone is building a new iOS healthcare app (and thousands are), what standard do we point them at?
- If someone wants to provide a cloud based health app that integrates with social networks, what standard should they use?

Requirements for Stage 2 Meaningful Use

An Instructional Webinar Series

- February 20 & 27 – Consolidated CDA Implementation Guide, Part 1 & 2
- March 6 –Family Health History and Beyond
- March 13 – S&I Framework Laboratory Result Interface Implementation Guide Using HL7 Version 2.5.1
- March 20 – Immunization Messaging Using HL7 Version 2.5.1
- March 27 – Electronic Laboratory Reporting to Public Health Using HL7 Version 2.5.1
- April 3 – Infobuttons for Clinical Decision Support
- April 10 – HL7 Quality Reporting Document Architecture (QRDA)

Standards and Select IP Freely Available

Contact:
Andrea Ribick
+1 (734) 677-7777
andrea@HL7.org

HL7 Standards Soon to be Free of Charge

Health IT Standards Leader to Support Widespread Global Adoption by Making Standards and Select IP Freely Available

Ann Arbor, Michigan, USA – Sept. 4, 2012 – Health Level Seven® International (HL7®), the global leader in developing interoperability standards for healthcare IT, announced today its decision to make much of its intellectual property (IP), including standards, freely available under licensing terms. The landmark decision represents HL7's commitment to the betterment of healthcare worldwide by ensuring that all stakeholders have equal access to its HIT standards. The new policy is expected to take effect in the first quarter of 2013.

New Member Benefits

How to get more info on HL7

- Web site:
  - http://www.hl7.org
- International Affiliates
  - http://www.hl7.org/Special/committees/international/intl.htm
- Education and Tutorials
  - http://www.hl7.org/education/index.cfm
- How to request and HL7 Ambassador speaker
  - mailto:hq@hl7.org
- Contact info for HL7 HQ
  - mailto:hq@hl7.org
- Product and Services Guide
  - http://productsandservices.hl7.org/Report/Report.aspx?varReport=Product

Thank You

The HL7 Healthcare Connection

An HL7 Overview

HL7 Ambassador Webinar

January 30, 2013

Grant M. Wood

Intermountain Healthcare Clinical Genetics Institute

and HL7 Clinical Genomics workgroup