Preparing for the Collection of External Family History & Genetic Test Result Data

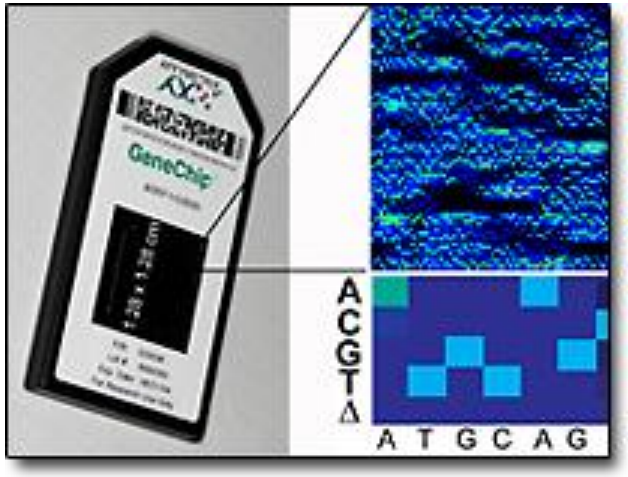
HL7 Ambassador Webinar

Grant M. Wood

Clinical Genomics Workgroup

October 3, 2012

Genetic Testing Becoming Common

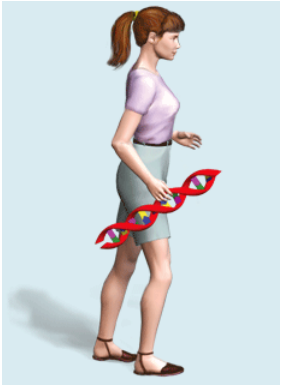


The good news from human genome research is that tests to determine people's genetic susceptibility to many common and deadly diseases are already, or soon will be, available.

Are Healthcare Systems Prepared

The bad news is that most health care systems risk being overwhelmed unless they start preparing for the complex and costly demands of genetic screening programs

A Personalized Medicine Story



A mother is concerned about a family history of breast cancer or cardiovascular disease

She completes one of the new online programs that collect family health history

- ➲ And takes the paper copy to her doctor
- ➲ And makes her husband also take a copy to his doctor

The question is –

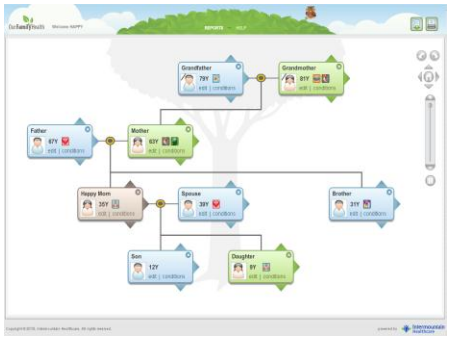
Will the family health history data ever be stored and shared electronically with other healthcare providers?

Will the data be available to help their children? Grandchildren?

A Personalized Medicine Story

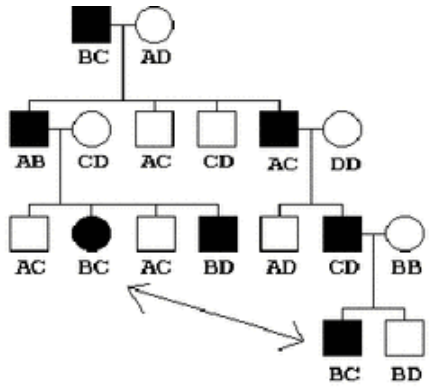
They have many choices to enter information –

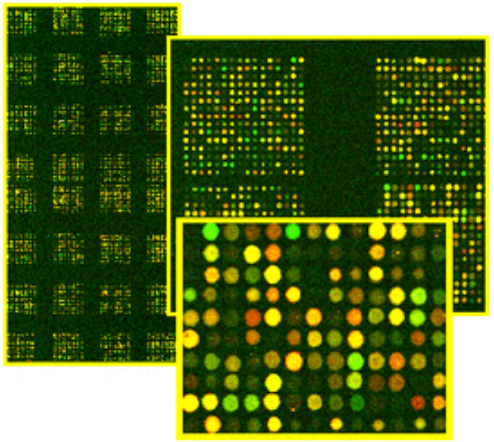
1. In an employer, health plan, or healthcare system Personal Health Record or Patient Portal
2. Or they use a family history tool on one of the new commercial PHRs
3. They do genealogy online. Shouldn't they also do family health history at the same?



Will this data be recorded in the Electronic Health Record (EHR) of their healthcare provider?

Value of Family History in Clinical Care





Family history remains the best and least expensive genetic 'test' currently available for clinical use.

A major effort will entail developing tools to collect this information –

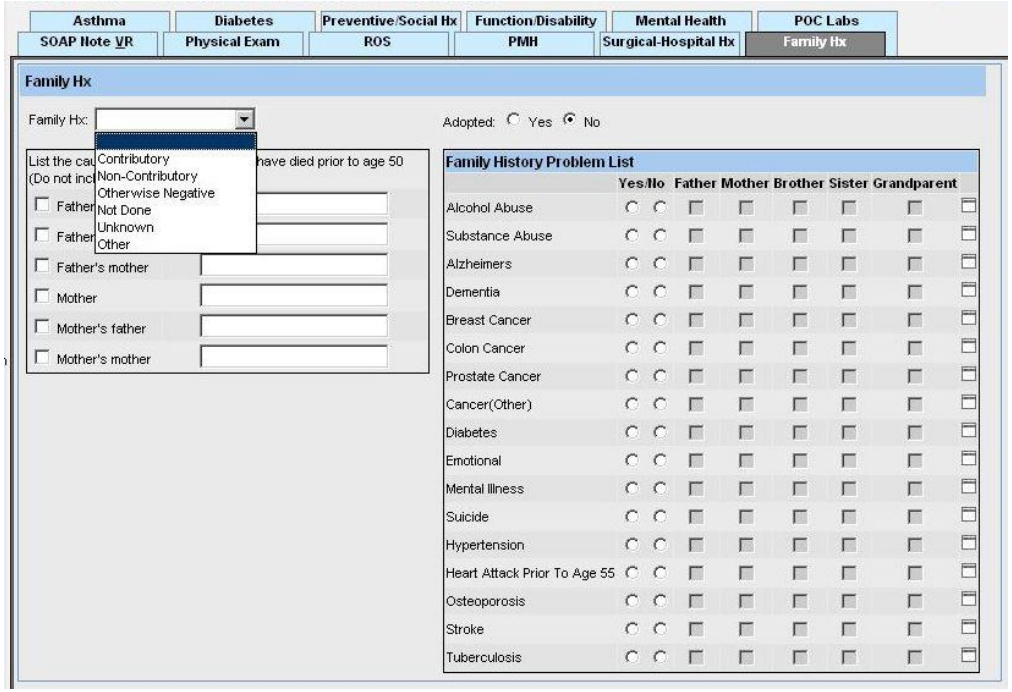
1. In a standardized format,
2. Store it in the patient's electronic health record,
3. Apply risk assessment, and
4. Develop messages to clinicians that may alter patient care based on the information obtained.

Current Methods of Collecting Data

Text-based data

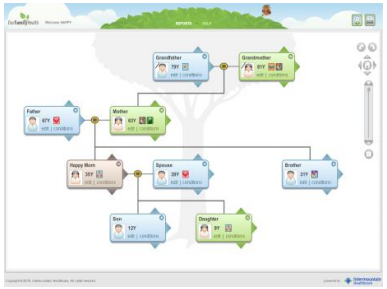
- "FAMILY HISTORY: positive for diabetes, end-stage renal disease requiring hemodialysis in her father and mother, and multiple siblings have a history of coronary artery disease."
- "...father died at age 40 of sudden cardiac death from myocardial infarction. Mother died at age 56 with MI. He notes that 15 people have died of coronary artery disease in the last three generations of his family. Diabetes type II in multiple members of the family. Denies cancer, seizures, or hyperlipidemia."

An electronic form, but still insufficient



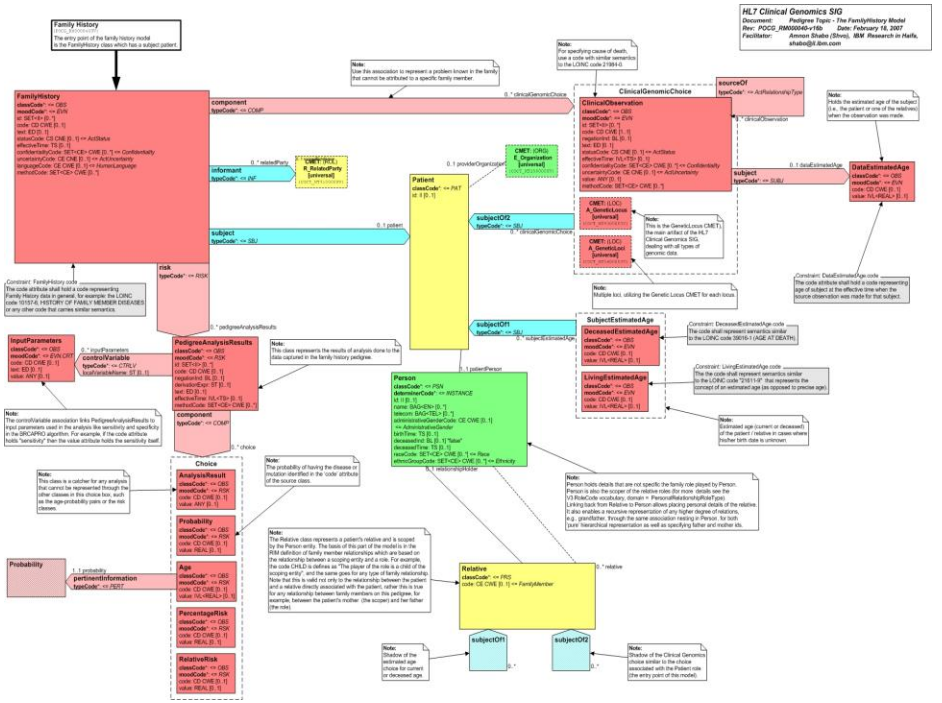
HL7 Family History Model (Pedigree)

- The XML-based model can be used for family health history data storage in an EHR.
- It is fundamentally designed for -
  1. The interoperability of family history data between both patient entered systems and clinical information systems,
  2. And provide structured data for risk analysis and clinical decision support.

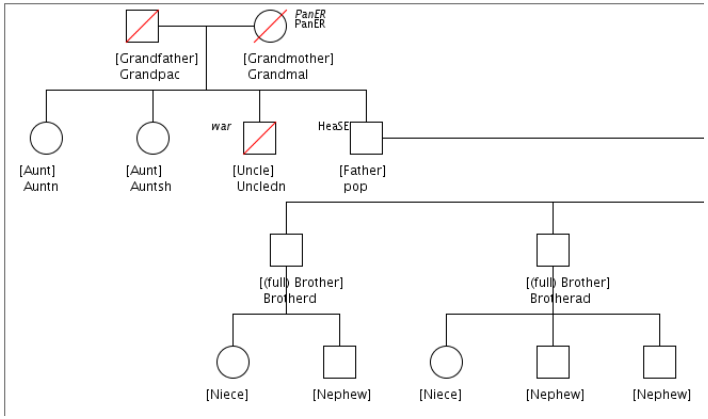


What's In the Family History Data Model

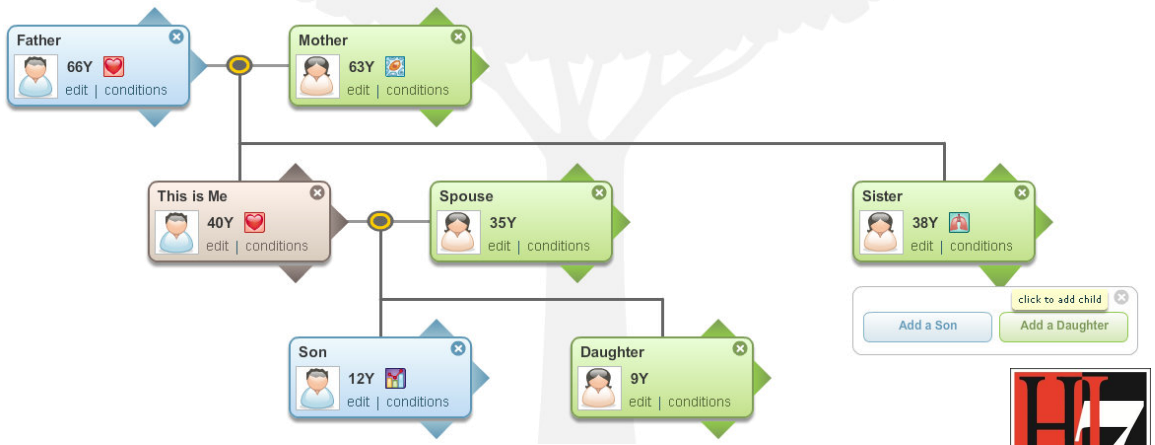
1) Record information
2) Person of focus (Proband)
3) Other persons in pedigree
4) Age of person / death date
5) Relationship
6) Disease
7) Age of disease onset / age of disease death
8) Genotypic data
9) Risk analysis



Data That Can Be Transmitted



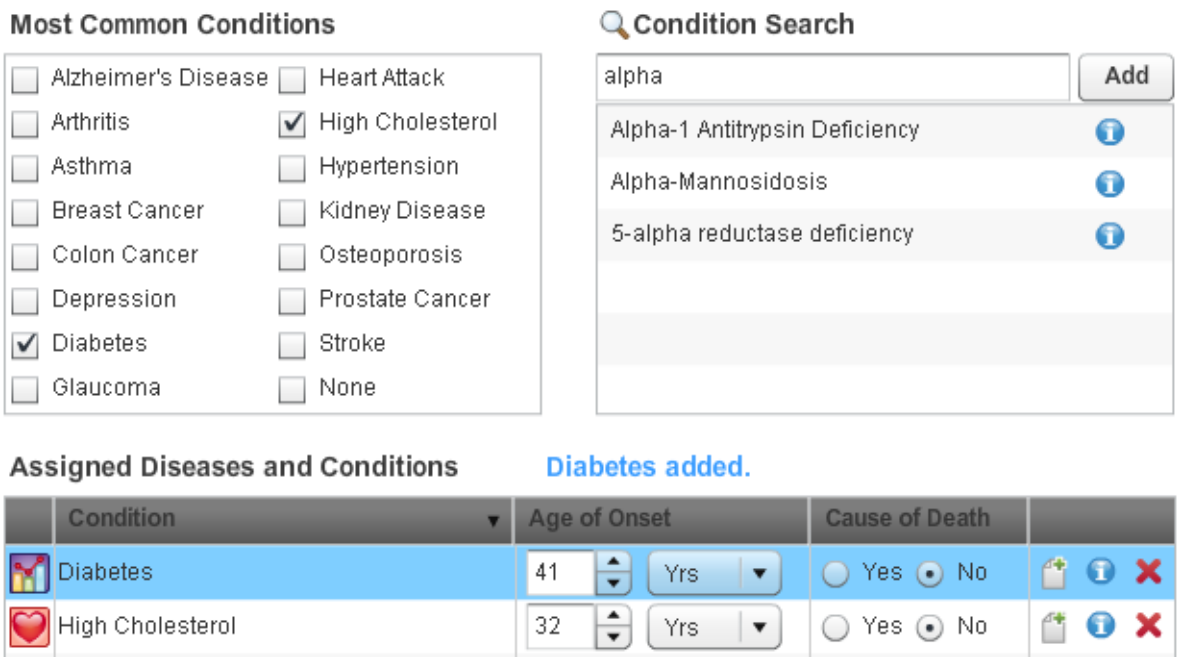
Full pedigree data from one application and completely re-drawn in another



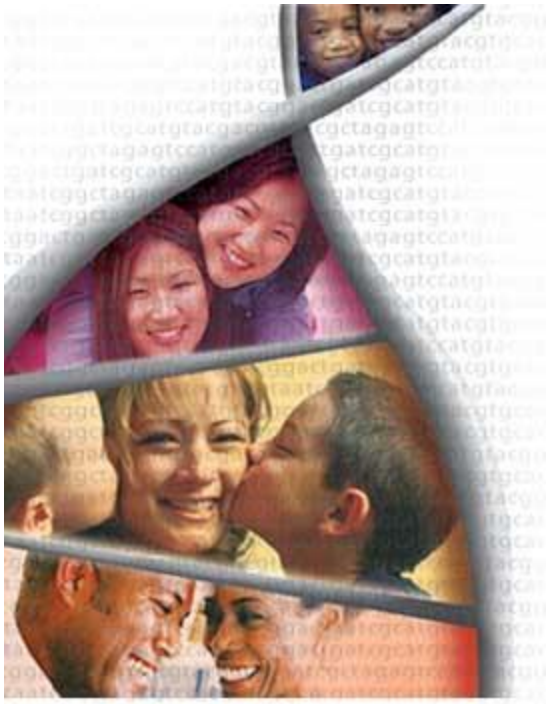
Data That Can Be Transmitted

Clinical Data

1. Disease or condition for each relative, using SNOMED or other coding system
2. Age of Onset
3. Age of Death
4. Cause of Death



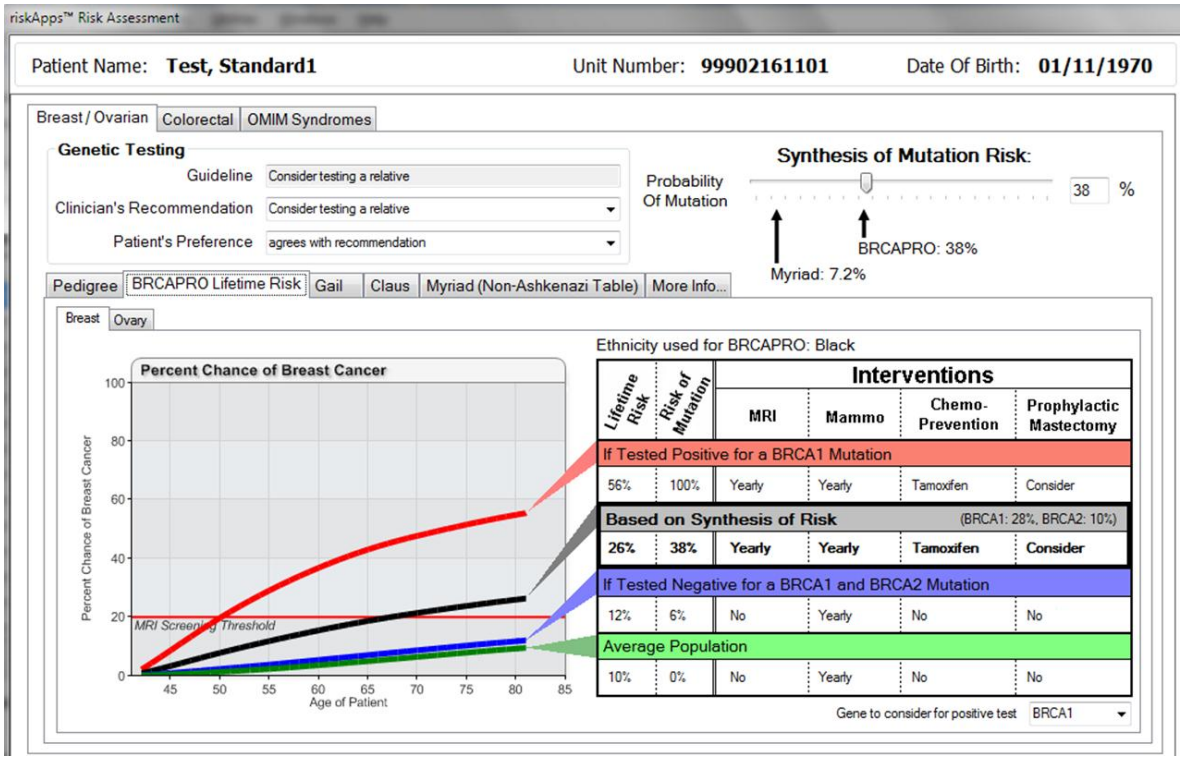
Data That Can Be Transmitted



Genetic Test Results in XML

```
<!-- GENOMIC DATA  -->
  <subjectOf2>
  <geneticLocus moodCode="EVN">
  <component1>
  <individualAllele moodCode="EVN">
  <text>breast cancer 2, early onset</text>
  <value code="U43746" displayName="BRCA2"
codeSystemName="HUGO" />
  <component3>
  <sequenceVariation moodCode="EVN">
  <value xsi:type="CE" code="185delAG" />
  <interpretationCode code="DELETERIOUS" />
```

Data That Can Be Transmitted



Screen shot from Hughes riskApps™

Risk Analysis

1. Risk scoring calculated by advanced programs can be shared.
2. Disease-specific risk algorithms can be provided by web services.

My Family Health Portrait

A tool from the Surgeon General

Using *My Family Health Portrait* you can:

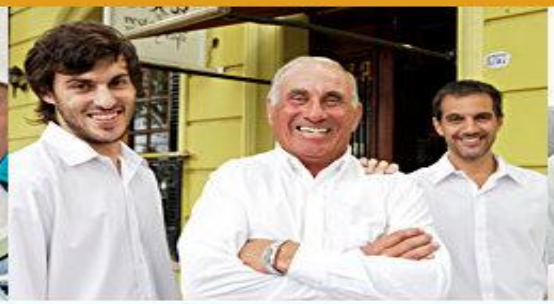
- Enter your family health history.
- Create drawings of your family health history to share with family or health care worker.
- Use the health history of your family to create your own.

Talking with your health care worker about your family health history can help you stay healthy!

Learn more about *My Family Health Portrait*

Create a Family History

Open a Saved History File

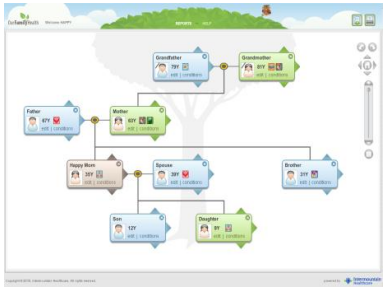



Meaningful Use Stage 2 Requirement For Family History

M4	**Patient Family Health History**	**Record patient family health history as structured data**	**More than 20% of all unique patients admitted to the eligible hospital or CAH's inpatient or emergency department during the EHR reporting period have a structured data entry for one or more first-degree relatives or an indication that family health history has been reviewed**

Invitation and Challenge

Since becoming a normative HL7 standard in 2007, the family history model has also been approved as an ANSI standard. HL7 Clinical Genomics is now calling on all commercial vendors and those who have developed “in-house” systems to implement this data model and electronic messaging standard in their clinical products and applications.



Going Beyond Family History

A Personalized Medicine Story

They spend money on the commercially available direct-to-consumer genetic testing

OR

She has a BRCA gene breast cancer test ordered by her doctor

HL7 Genetic Variation Data Model

The Genetic Variation model specifies the structure and semantics for the transmission of information created during single or multiple gene testing.

The model is further constrained to genetic variation analyses based upon sequence variation, and derived from a set of scientific laboratory methods, such as:

1. **SNP probes,**
2. **Genotyping,**
3. **and Gene Sequencing**

that focus on genetic changes, usually in the coding region(s) of one or a small number of genes.

HL7 Genetic Variation Data Model

The model facilitates the electronic transmission of genetic testing results and interpretations from –

- Genetic testing laboratories to medical practitioners, electronic health records, personal health records and associated clinical decision support systems able to receive and process such information
- Genetic testing laboratories to drug and medical device companies that have ordered such information as part of a clinical trial
- Drug and medical device companies to regulatory agencies that need to review such information as part of a new drug or device marketing application

V2_CG_LOINCGENVAR_R2_INFORM_2011DEC

HL7 Version 2 Implementation Guide: Clinical Genomics; Fully LOINC-Qualified Genetic Variation Model, Release 2

Submitted for Ballot December, 2011

HL7 Informative Document for Ballot

Sponsored by:

Clinical Genomics WG

Principal Contributors:

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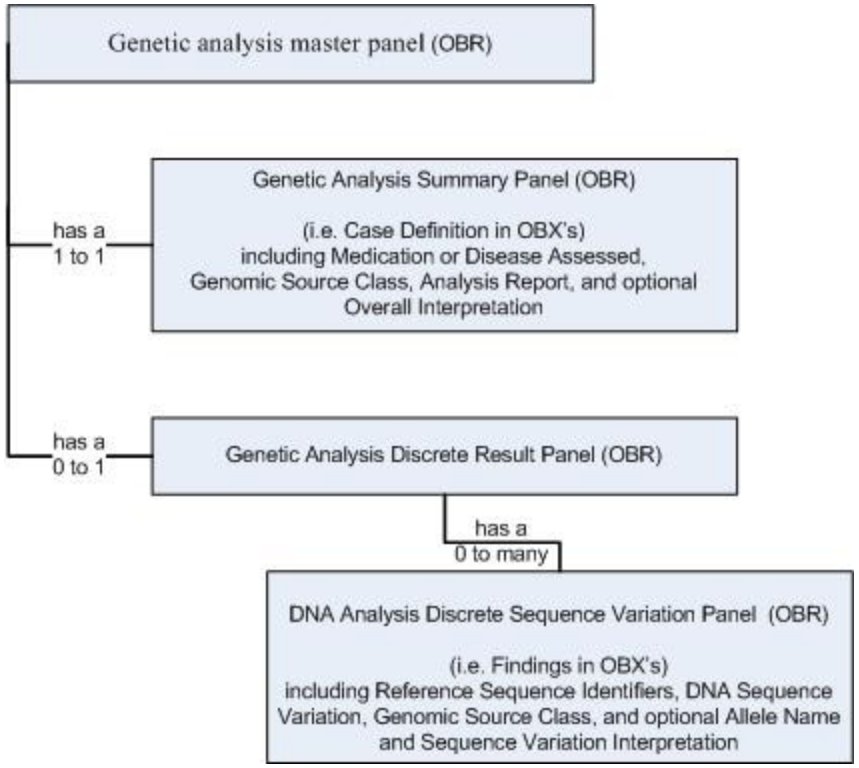
Grant Wood
Stan Huff
Clement McDonald

Genetic Variation Implementation Guide



Genetic Variation Implementation Guide

Nomenclatures, Code Systems and Value Sets

- LOINC
- SNOMED
- RxNORM
- HGNC
- HGVS
- DbSNP
- RefSeq

Genetic Variation Implementation Guide

TABLE 7-2 -GENETIC ANALYSIS SUMMARY PANEL

OBR/ OBX	OBX-2 Value Type	Usage*	Card-inality	Value Set	LOINC Code	LOINC Element Name	Description/Comments
OBR		R	1..n		55232-3	Genetic Analysis Summary Panel	The summary panel for a genetic analysis for one or more laboratory tests (e.g. analysis for disease risk, diagnosis or pharmacogenetics) on a single accession.
OBX	CWE	C1		SNOMED	51967-8	Genetic disease assessed	A coded disease (recommend SNOMED) which is associated with the region of DNA covered by the genetic test.
OBX	CWE	C1		RxNORM	51963-7	Medication Assessed	A coded medication accessed in a pharmacogenic test (recommend RxNorm).
OBX	CWE	R			48002-0	Genomic Source Class	The genomic class of the specimen being analyzed: Germline for inherited genome, somatic for cancer genome (e.g. DNA from tumor cells), and prenatal for fetal genome. LOINC Answer List values can be seen in Table 7.6.

If the study is intended to assess disease risk or diagnosis based on genetic findings, then the *Genetic Disease Analysis Overall Interpretation* is used (see below).

Genetic Variation Implementation Guide

LOINC #	Component Name
47998-0	DNA Sequence Variation display name
47999-8	DNA region name
48002-0	Genomic source class
48003-8	DNA Sequence Variation identifier
48004-6	DNA Sequence Variation
48005-3	Amino acid change
48006-1	Amino acid change type
48008-7	Allele name
48013-7	Genomic reference sequence identifier
48018-6	Gene identifier
48019-4	DNA Sequence Variation type
51958-7	Transcript reference sequence identifier
51959-5	DNA region of interest
51961-1	Drug efficacy sequence variation interpretation
51963-7	Medication assessed
51964-5	Drug efficacy analysis overall interpretation
51967-8	Genetic disease assessed
51968-6	Genetic Disease Analysis Overall Interpretation
51969-4	Genetic analysis summary report
51971-0	Drug metabolism analysis overall interpretation
53034-5	Allelic state
53037-8	Genetic disease sequence variation interpretation
53039-4	Genetic disease analysis overall carrier interpretation
53040-2	Drug metabolism sequence variation interpretation

Genetic Variation Implementation Guide

TABLE 7-6 – LOINC ANSWER LISTS

LOINC code	LOINC component	Sequence	Answer text	LOINC answer code
53034-5	Allelic state	1	Heteroplasmic	LA6703-8
		2	Homoplasmic	LA6704-6
		3	Homozygous	LA6705-3
		4	Heterozygous	LA6706-1
		5	Hemizygous	LA6707-9
48006-1	Amino acid change type	1	Wild type	LA9658-1
		2	Deletion	LA6692-3
		3	Duplication	LA6686-5
		4	Frameshift	LA6694-9
		5	Initiating Methionine	LA6695-6
		6	Insertion	LA6687-3
		7	Insertion and Deletion	LA9659-9
		8	Missense	LA6698-0
		9	Nonsense	LA6699-8
		10	Silent	LA6700-4
		11	Stop Codon Mutation	LA6701-2
48019-4	DNA sequence variation type	1	Wild type	LA9658-1
		2	Deletion	LA6692-3
		3	Duplication	LA6686-5
		4	Insertion	LA6687-3
		5	Insertion/Deletion	LA6688-1
		6	Inversion	LA6689-9
		7	Substitution	LA6690-7
51964-5	Drug efficacy analysis overall interpretation	1	Responsive	LA6677-4
		2	Resistant	LA6676-6
		3	Negative	LA6577-6
		4	Inconclusive	LA9663-1
		5	Failure	LA9664-9
51961-1	Drug efficacy sequence variation interpretation	1	Resistant	LA6676-6
		2	Responsive	LA6677-4
		3	Presumed resistant	LA9660-7

HL7 Version 2 Message

Genetic Disease Analysis

8.2.1 Example: Genetic Disease Analysis (e.g. Dilated Cardiomyopathy)
MSH-->As according to HL7 VERSION 2.5.1 IMPLEMENTATION GUIDE: ORDERS AND OBSERVATIONS; INTEROPERABLE LABORATORY RESULT REPORTING TO EHR (US REALM), RELEASE 1, ORU^R01, HL7 Version 2.5.1, November, 2007.

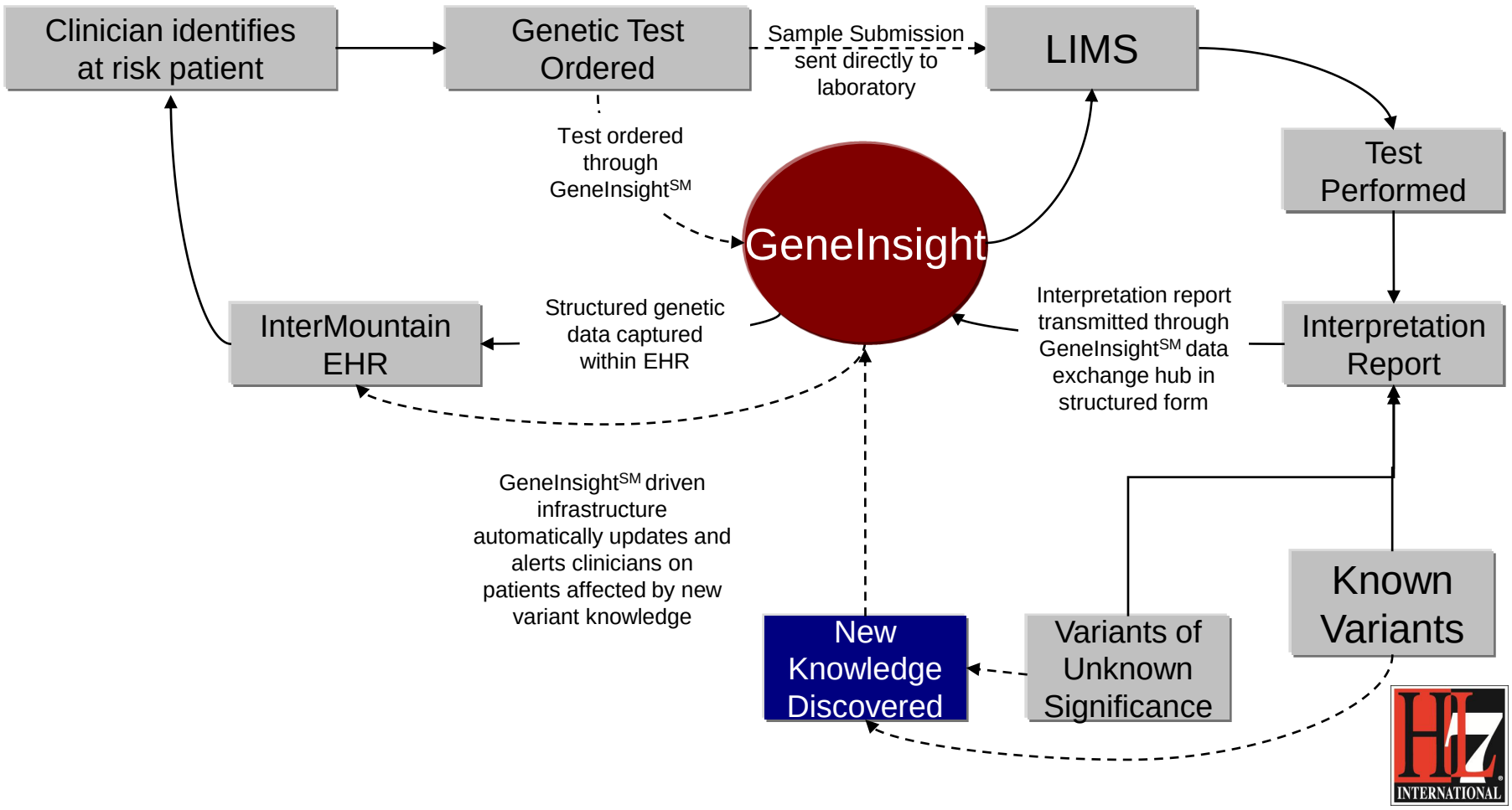
OBR|1||PM-08-J00094^HPCGG-LMM^2.16.840.1.113883.3.167.1^ISO|lm_DCM-pnlB_L^**Dilated Cardiomyopathy Panel B (5 genes)**^99LMM-ORDER-TEST-ID||20080702000000|20080702100909|||||||234567891^Pump^Patrick^^^^^^NPI^L|||||20080703000000|||F|||||00000009^Cardiovascular^99HPCGG-GVIE-INDICATION^^^^^^Clinical Diagnosis and Family History of DCM|&Geneticist&Gene&&&&&NPI^^^^^^^HPCGG-LMM&2.16.840.1.113883.3.167.1&ISO|||||||||||||55233-1^**Genetic analysis master panel** ^LN

SPM|1|||119273009&Peripheral blood&SNM3&&&&0707Intl&&Blood, Peripheral||||||||||||20080702000000

OBR|2||PM-08-J00094-1^HPCGG-LMM^2.16.840.1.113883.3.167.1^ISO|55232-3^**Genetic analysis summary panel**^LN|||20080702000000|||||||||||||20080703000000|||F||||^PM-08-J00094&HPCGG-LMM&2.16.840.1.113883.3.167.1&ISO

OBX|1|CWE|51967-8^**Genetic disease assessed**^LN||399020009^DCM-Dilated Cardiomyopathy^SNM3^^^0707Intl||||||F|20080702100909||||||||||Laboratory for Molecular Medicine^L^22D1005307^^^CLIA&2.16.840.1.113883.4.7&ISO|1000 Laboratory Lane^Ste. 123^Cambridge^MA^99999^USA^B

Advanced Genetic Testing Workflow



Other Areas of Activity

- CDA template – for CDA-based transfer of genetic test results
- Cytogenetics – for chromosomal-based tests in the clinic
- Gene Expression – for laboratory research and drug discovery
- Whole Genome Sequencing – rapid advancing next generation sequencing technology

Implementation Guide for CDA Release 2
Genetic Testing Report (GTR)
(Universal Realm)

Draft For Comment

Informal Review

May 2012

CDAR2_IG_GENTESTRPT_R1

CDA is the Basis For ...

- Continuity of Care Document
- Consult Note
- 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 …

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"?>
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 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.

HL7 VERSION 2 IMPLEMENTATION GUIDE: CLINICAL GENOMICS; FULLY LOINC-QUALIFIED CYTOGENETICS MODEL, RELEASE 1

ORU^R01

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The hierarchical structures of panels are shown in the following diagrams:

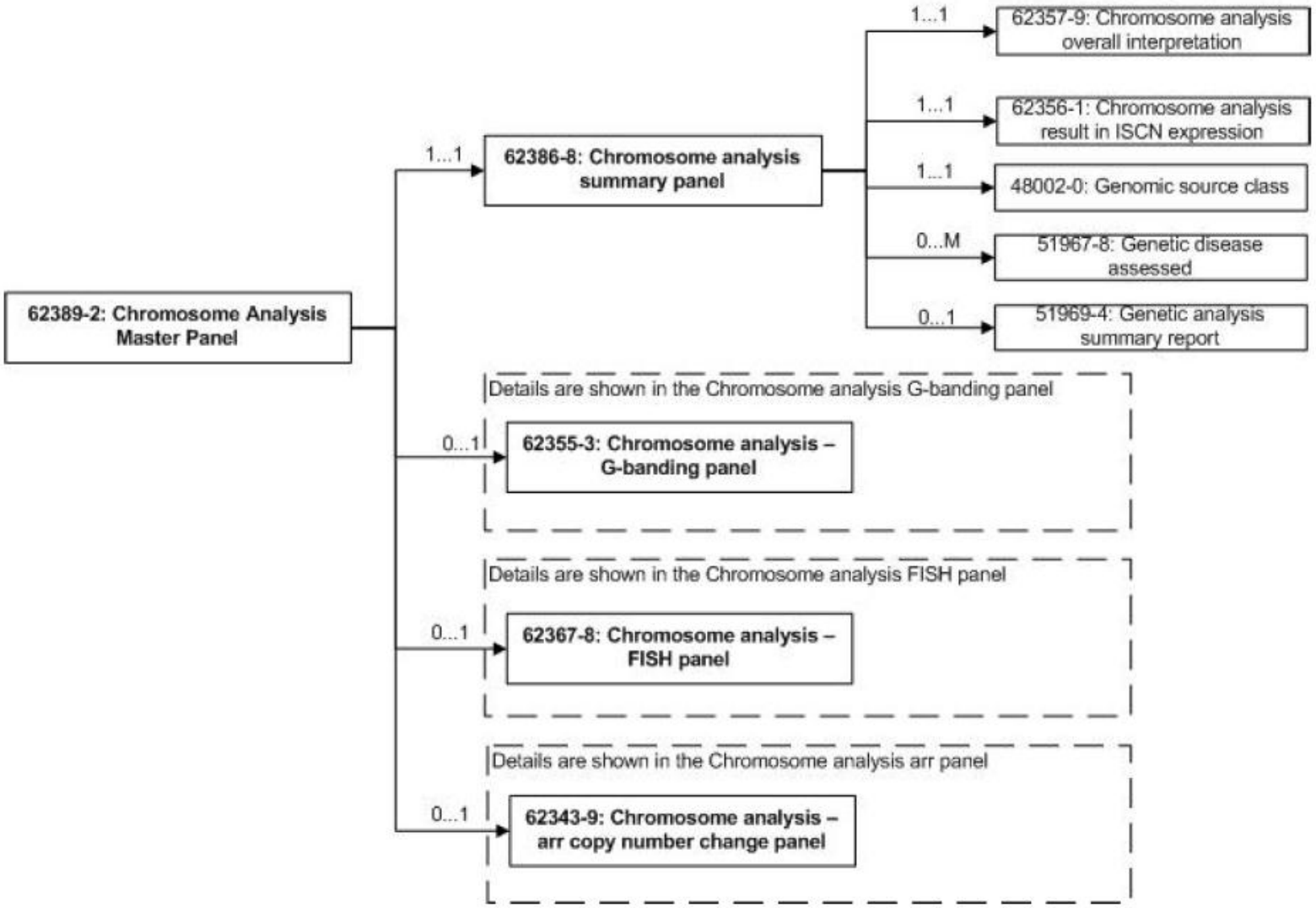

Figure 1: Chromosome analysis master panel

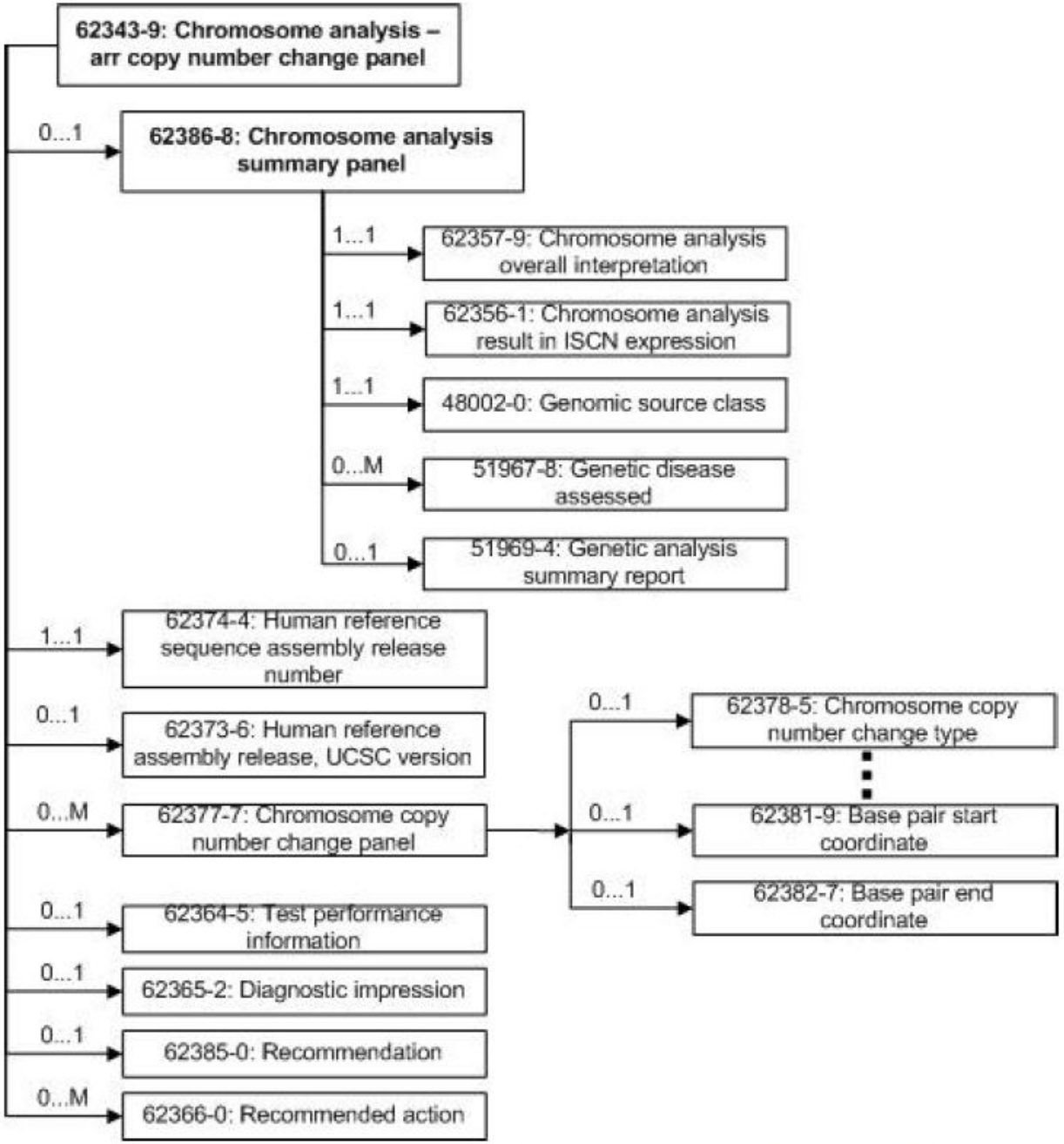

Figure 4: Chromosome analysis arr copy number change panel

Personalized Medicine Use Case

Scientists find genes that could predict Type 2 diabetes

International scientists identified five different genetic variations tied to adult-onset diabetes and believed to be responsible for 70% of the genetic risk for the diabetes, also known as Type 2. One of the lead scientists says the findings "mean we can create a good genetic test to predict people's risk of developing this type of diabetes.“

1. Family history risk assessment
2. Order genetic test
3. Test interpretation
4. Store results (family health history, sequence data, alleles, exons, SNP’s also called variations or mutations)
5. Clinical decision support
6. Pharmacogenomics for targeted drugs

Adding to the List

- V3 Family Health History model
- V2 Genetic Variation model
- CDA Genetic Test Result electronic document
- V2 Cytogenetics model
- Gene Expression – for laboratory research and drug discovery
- **Next Generation Sequencing**

Stakeholder Landscape

- Vendors
  - Life Technologies: SOLiD 4, 5500
  - Ion Torrent: PGM
  - Illumina: HiSeq, MiSeq
  - Complete Genomics
  - Pacific Biosciences
- Consumers
  - Clinical Reference Labs
  - Academic Clinical labs
  - Hospital Labs
- Other Stakeholders
  - FDA, NIST, CAP

Detailed Use Cases to drive workflow from order to report

- Rooted in existing technologies
  - Genome Analysis (whole genome, whole exome)
  - SNP Genotyping and genetic rearrangements
  - Multiplex gene panels or arrays (CFTR, array CGH, cancer panels)
  - Targeted gene sequencing (developmental delay, cardiomyopathy)

Detailed Use Cases to drive workflow from order to report

- Near term use cases
  - Cancer (risk assessment, somatic mutations, low frequency)
  - Germline / Constitutional (trios, pedigrees)
  - Infectious Diseases (viral, bacterial)
  - Re-interpretation of existing data
  - Research (microbiome, immunome)

Invitation and Challenge

HL7 Clinical Genomics is also calling on forward thinking organizations to test and comment on Genetic/Genomic/Sequencing models, as we work towards extending the standard.

Contact Information

- Find the HL7 Clinical Genomics workgroup at –
  - http://www.hl7.org/Special/committees/clingenomics/index.cfm
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Questions