

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

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1. Introduction

The *HL7 VERSION 2 IMPLEMENTATION GUIDE: CLINICAL GENOMICS; FULLY LOINC-QUALIFIED CYTOGENETICS MODEL, RELEASE 1* details structuring cytogenetics test results into the electronic health record utilizing HL7 version 2.5.1. This implementation guide is modeled after the *HL7 Version 2.5.1 Implementation Guide: Orders And Observations; Interoperable Laboratory Result Reporting To EHR (US Realm), Release 1*, and covers the reporting of cytogenetics results for constitutional cytogenetic tests.

Genetic testing is rapidly changing the way we practice medicine. According to GeneTests.org, the number of genetic tests for disease is now over 2,300, a significant increase from 110 that was first tracked in 1993. The number of clinically available tests went up 300 in the past two years alone. When you combine the physician's lack of basic knowledge and confidence about clinical genomics with the vast quantity and complexity of genetic and genomic data, that opens up an opportunity for health information technology, such as clinical decision support. As stated by U.S. Department of Health & Human Services on Personalized Health Care, the growing base of biomedical knowledge (especially related to genomic knowledge) and the adoption of interoperable health information technology are the building blocks to the achievement of personalized health care.¹

Cytogenetics is a specialty of genetic testing. Cytogenetics evaluates whole chromosomes from the nucleus of the cell for changes in number or structure.² Both traditional and molecular techniques can be applied to cytogenetic testing. Conventional banding uses a microscope to visually examine whole genomes. Molecular cytogenetic methods such as Fluorescence In Situ Hybridization (FISH) and cytogenomic microarray (arr) allow cytogeneticists to examine the genome at a greater resolution. The FISH technique uses individual probes to reveal DNA gains and losses, or rearrangements of the probe-targeted segments, but does not provide any information about the rest of the genome.³ The development of cytogenetic microarray overcomes this limitation. It allows for genome wide detection of DNA sequence copy number changes. The resolution of traditional banding technique is limited to the detection of genomic imbalances in the 5-10 Mb range. The resolution of FISH far exceeds conventional banding technique. It detects submicroscopic changes to 50 kb in a clinical cytogenetic setting. Oligonucleotide platforms for microarray analysis can detect genomic imbalances as small as a 500 bp.

The complexity of genetic data requires additional coding of the message components using LOINC. These codes are listed in tables in Section 7. LOINC coding has several advantages, including more robust representation of the data when persisted in a database, increased accuracy when supporting multiple HL7 message formats, and consistency of representation for clinical decision support.

The chapters in this guide that describe messaging infrastructure, abstract message syntax, and segment and field descriptions are based on chapters from the parent implementation guide entitled **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**. This guide can be found at http://www.hl7.org/Memonly/downloads/Standards_Messaging_V251/InteroperabilitySpecificationLabResultMessage_v251.zip (HL7 membership required).

¹ <http://www.hhs.gov/myhealthcare/news/personalized-healthcare-9-2007.html#Pathways>

² http://www.medscape.com/viewarticle/505220_6

³ <http://www.aacc.org/publications/cln/2010/may/Pages/SeriesArticle.aspx>

1.1 PURPOSE

The *HL7 VERSION 2 IMPLEMENTATION GUIDE: CLINICAL GENOMICS; FULLY LOINC-QUALIFIED CYTOGENETICS MODEL, RELEASE 1* is a message specification intended to standardize the electronic reporting of cytogenetic test results from clinical laboratories to Electronic Health Record (EHR) systems. It is specific to the US Realm. This implementation guide will specify the use of LOINC panels for cytogenetics and its associated codes to transmit coded and structured cytogenetic testing result reports in HL7 version 2 messages. Note these codes may also be used in version 3 messages. As an example, a current HL7 project illustrates this use: see the document *Implementation Guide for CDA Release 2 Genetic Testing Report, Universal Realm*.

1.2 AUDIENCE

This guide is designed to be used by analysts and developers who require guidance on the reporting of cytogenetics results using standard LOINC codes and in a structured format.

1.3 SCOPE

There are two types of cytogenetic abnormality. In the first type, a constitutional (i.e. germline) cytogenetic abnormality is typically present in nearly all nuclei-containing cells of the patient. In the second type, an acquired (i.e., somatic) cytogenetic abnormality represents a genetic change, for example a cancerous change associated with a neoplastic process. This guide focuses on the reporting of constitutional cytogenetic test results. It covers constitutional cytogenetic test results performed using conventional banding, FISH, and arr for copy number changes techniques.

Constitutional cytogenetics refers to the study of the molecular aspects of heredity, chromosome structure, and the identification of genetic aberrations and variants linked to disease. Note constitutional is used in the field of Cytogenetics and germline within molecular diagnostics (e.g. sequence variations). For our purposes these terms can be assumed equivalent. Cytogenomic SNP microarray (for identification of DNA sequence variations in the sequence of the genome) is not in the scope of this release.⁴ In addition, cancerous cytogenetic test result reports are not in the scope of this release. However, it is believed that this message will readily scale to support cancerous cytogenetic testing.

Finally, LOINC answer lists for Genomic Source Class is used within this implementation guide and may be used to classify findings and testing scenarios from germline, prenatal, or somatic (for tumor testing).

- **Use of Vocabulary Standards** 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.

⁴ [http://www.aruplab.com/Testing-Information/resources/TechnicalBulletins/November2010/Cytogenomic%20SNP%20Microarray%20\(CMASNP\).pdf](http://www.aruplab.com/Testing-Information/resources/TechnicalBulletins/November2010/Cytogenomic%20SNP%20Microarray%20(CMASNP).pdf)

1.4 ASSUMPTIONS

Assumptions are summarized as follows:

- Infrastructure is in place to allow accurate information exchange between information systems.
- Providers access lab test results through either an EHR or a clinical data system.
- Privacy and security has been implemented at an acceptable level.
- All participants agree to all standards, methodologies, consent, privacy and security.
- Legal and governance issues regarding data access authorizations, data ownership and data use are outside the scope of this document.
- The order, paper or electronic, associated with the lab result contains sufficient information for the laboratory to construct the lab result message properly.

1.5 CONVENTIONS

The following conventions have been used in establishing this guide:

- The rules outlined in *HL7 2.5.1, Chapter 2, Section 2.12, Conformance Using Message Profiles*, were used to document the use case for, and constraints applied to, the messages described in this guide.
- Data types have been described separately from the fields that use the data types. For details regarding data type field lengths, please refer to *Section 2.1.3, Lengths*, in this document.

2. Messaging Infrastructure

The V2 Cytogenetics model uses the same messaging infrastructure as described in Chapter 2, Page 3 of the parent implementation guide entitled **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.** The guide can be found at http://www.hl7.org/Memonly/downloads/Standards_Messaging_V251/InteroperabilitySpecificationLabRes ultMessage_v251.zip (HL7 membership required).

3. Message Profile – Genetic Laboratory to EHR

3.1 USE CASE MODEL

TABLE 3-1 – USE CASE LABORATORY TO EHR

Description	Clinical applications of cytogenetic testing include assessment of a developmentally delayed child, evaluation of a cancerous tumor, or amniocentesis to detect chromosomal anomalies in a fetus. This implementation guide covers reporting of constitutional cytogenetic test results from clinical laboratories to EHR systems. The reporting of cancer cytogenetics will be covered in the future releases.
Actors	<p>Laboratory Result Sender – The laboratory result sender actor is an application capable of performing laboratory testing on specimens. The laboratory application is capable of transmitting the results of laboratory testing to a receiver. In the use case, the laboratory result sender is identified as a "Laboratory Organization."</p> <p>Laboratory Result Receiver – The laboratory result receiver is an application capable of receiving results of laboratory testing. Typically this actor represents an EHR application. The laboratory result receiver may be associated with the ordering provider or another provider, commonly referred to as a "copy-to provider," that needs to have access to the results. In the use case, the laboratory result receiver is identified as either the "Clinician" or "Data Repository."</p>
Assumptions	<p>Assumptions are summarized as follows:</p> <ul style="list-style-type: none"> Infrastructure is in place to allow correct information exchange between information systems. Providers access lab test results either through an EHR or a clinical data system. Privacy and security has been implemented at an acceptable level. All participants agree to all standards, methodologies, consent, privacy and security. Legal and governance issues regarding data access authorizations, data ownership and data use are outside the scope of this document. <p>The following are preconditions for the use of this profile:</p> <ul style="list-style-type: none"> The order contains the unambiguous names and electronic addresses for the other authorized providers of care. When needed, the patient is registered in a Patient ID Cross-Referencing system that includes both the laboratory patient ID and the clinician's patient ID. For the electronic laboratory result, the laboratory has transformed any local codes into HITSP-specified terminologies before transmission. <p>Additional Preconditions:</p> <ul style="list-style-type: none"> A valid order for laboratory testing exists.

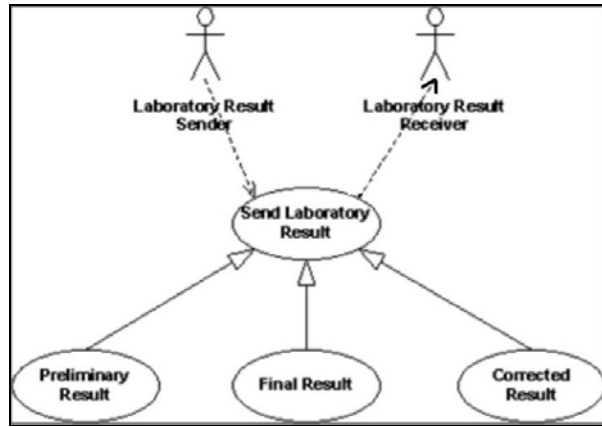


Figure 3-1. Send Genetic Laboratory Result Use Case Model

3.2 DYNAMIC INTERACTION MODEL

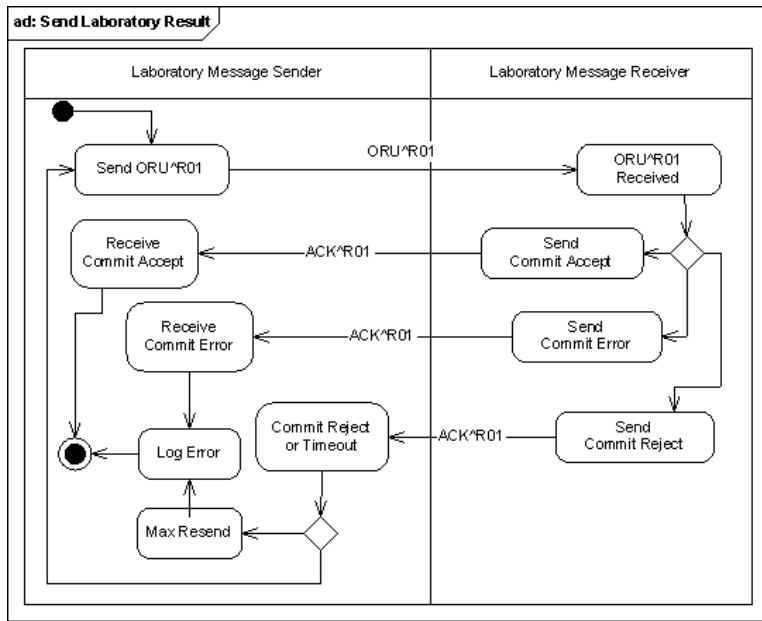


Figure 3-2. Activity Diagram for Send Genetic Laboratory Result Use Case

3.3 DYNAMIC DEFINITION

TABLE 3-2 – DYNAMIC DEFINITION	
Item	Value
Profile ID	USLabReport
HL7 Version	2.5.1
Accept Acknowledgement	AL – Always
Application Acknowledgement	For valid values, refer to HL7 Table 0155 – Accept/Application Acknowledgment conditions in section 5.2.1 in 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. The guide can be found at http://www.hl7.org/Memonly/downloads/Standards_Messaging_V251/InteroperabilitySpecificationLabResultMessage_v251.zip (HL7 membership required). .
Acknowledgement Mode	Immediate
Profile Type	Realm Constrainable Profile
Message Types	ORU^R01^ORU_R01, ACK^R01^ACK
Encoding	ER7 (required) 2.5.1 XML (optional)

3.4 INTERACTIONS

TABLE 3-3 – INTERACTIONS							
Event	Description	Usage	When Used	Message Type	Receiver Action	Sender	Data Values
Order Received, No specimen	Order received; specimen not yet received	O	Preliminary Result	ORU^R01^ORU_R01	Commit Accept, Commit Reject or Commit Error	Laboratory Result Sender	ORC-1=RE OBR-25=O
Specimen Received	No results available; specimen received, procedure incomplete	O	Preliminary Result	ORU^R01^ORU_R01	Commit Accept, Commit Reject or Commit Error	Laboratory Result Sender	ORC-1=RE OBR-25=I
Procedure Scheduled	No results available; procedure scheduled, but not done	O	Preliminary Result	ORU^R01^ORU_R01	Commit Accept, Commit Reject or Commit Error	Laboratory Result Sender	ORC-1=RE OBR-25=S
Preliminary Result	Preliminary: A verified early result is available, final results not yet obtained	R	Preliminary Result	ORU^R01^ORU_R01	Commit Accept, Commit Reject or Commit Error	Laboratory Result Sender	ORC-1=RE OBR-25=P

TABLE 3-3 – INTERACTIONS

Event	Description	Usage	When Used	Message Type	Receiver Action	Sender	Data Values
Partial Result	Some, but not all, results available	O	Some Final Result	ORU^R01^ ORU_R01	Commit Accept, Commit Reject or Commit Error	Laboratory Result Sender	ORC-1=RE OBR-25=A
Unverified Result	Results stored; not yet verified	O	Preliminary Result	ORU^R01^ ORU_R01	Commit Accept, Commit Reject or Commit Error	Laboratory Result Sender	ORC-1=RE OBR-25=R
Final Result	Final results; results stored and verified. Can only be changed with a corrected result.	R	Final Result	ORU^R01^ ORU_R01	Commit Accept, Commit Reject or Commit Error	Laboratory Result Sender	ORC-1=RE OBR-25=F
Correction	Correction to results	R	Corrected Result	ORU^R01^ ORU_R01	Commit Accept, Commit Reject or Commit Error	Laboratory Result Sender	ORC-1=RE OBR-25=C
Testing Not Done	No results available; Order canceled.	O	Cancelled Test	ORU^R01^ ORU_R01	Commit Accept, Commit Reject or Commit Error	Laboratory Result Sender	ORC-1=RE OBR-25=X
No Order	No order on record for this test. (Used only on queries)	X	-	varies	NA	Laboratory Result Sender	ORC-1=RE OBR-25=Y
No Patient Record	No record of this patient. (Used only on queries)	X	-	varies	NA	Laboratory Result Sender	ORC-1=RE OBR-25=Z
Commit Accept	Enhanced mode: Accept acknowledgment : Commit Accept	R	All Cases	ACK^R01^ ACK	None	Laboratory Result Receiver	MSA-1=CA
Commit Error	Enhanced mode: Accept acknowledgment : Commit Error	R	All Cases	ACK^R01^ ACK	None	Laboratory Result Receiver	MSA-1=CE
Commit Reject	Enhanced mode: Accept acknowledgment : Commit Reject	R	All Cases	ACK^R01^ ACK	None	Laboratory Result Receiver	MSA-1=CR

4.Messages

The V2 Cytogenetics model uses the same messages as described in Chapter 4, Page 29 of the parent implementation guide entitled **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.** The guide can be found at http://www.hl7.org/Memonly/downloads/Standards_Messaging_V251/InteroperabilitySpecificationLabResultMessage_v251.zip (HL7 membership required).

5. Segment and Field Descriptions

The V2 Cytogenetics model uses the same segment and field descriptions as described in Chapter 5, Page 35 of the parent implementation guide entitled **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.** The guide can be found at http://www.hl7.org/Memonly/downloads/Standards_Messaging_V251/InteroperabilitySpecificationLabResultMessage_v251.zip (HL7 membership required).

6. Nomenclatures, Code Systems and Value Sets

6.1 VOCABULARY CONSTRAINTS

6.1.1 Genetic Tests, Testing Context, Interpretation Code, and Genetic Data

6.1.1.0 LOINC

TABLE 6-1 – LAB LOINC	
Code sets, vocabularies, terminologies and nomenclatures that need to be constrained	All LOINC lab result codes
Minimum attributes of the component:	HL7 value sets not established. Considered value sets should include: HEDIS (Health plan Employer Data and Information Set) reported tests accounting for 95% of routine lab orders. Proposed value sets for micro and cytology codes per HITSP/C80. Category A, B, & C bioterrorism agents/diseases. Public Health jurisdiction and Federal reportable disease conditions. The LOINC codes for reporting clinical genomics variation are listed in this implementation guide explicitly – and were developed and tested as described below.
Other Comments	LOINC - Vocabularies and code sets, useful in the reporting of genetic test result data into the EHR, in formats that can be leveraged by clinical decision support, have been defined as a result of the 2 year clinical pilot of the HL7 version 3 Genetic Variation model. These vocabularies and code sets have been submitted to LOINC and through ongoing collaborations between the National Library of Medicine's Lister Hill Center for Biomedical Communication, Partners HealthCare Center for Personalized Genetic Medicine (formerly the Harvard – Partners Center for Genetics and Genomics), Partners Healthcare, and Intermountain Healthcare, these vocabularies and codes will be piloted more broadly. In addition, the above collaborators have detailed these vocabularies and code sets in the HL7 implementation guide, balloted in Fall 2009, entitled: HL7 Version 2 Implementation Guide: Clinical Genomics; Fully LOINC-Qualified Genetic Variation Model, Release 1. The full LOINC data base can be downloaded from the Regenstrief Institute, Indianapolis, Indiana at http://loinc.org/ .

6.1.2 Associated Disease

6.1.2.0 SNOMED-CT

TABLE 6-2 – SNOMED-CT	
Code sets, vocabularies, terminologies and nomenclatures that need to be constrained:	SNOMED-CT
Minimum attributes of the component:	SNOMED-CT FDA SPL Problem List Subset
Other Comments:	FDA SPL Problem List Subset available at http://www.fda.gov/oc/datacouncil/term.html . The SNOMED terminology is used in the coding of disease associated with sequence variants or genes. Utilization of SNOMED provides linkage of genetic data with other clinical data stored in clinical applications.

6.1.3 Cytogenetics

6.1.3.0 ISCN

TABLE 6-3 – ISCN	
Code sets, vocabularies, terminologies and nomenclatures that need to be constrained:	The International System for Human Cytogenetic Nomenclature (ISCN)
Minimum attributes of the component:	ISCN nomenclature
OID	2.16.840.1.113883.6.299
Other Comments:	ISCN was created by the International Standing Committee on Human Cytogenetic Nomenclature to represent the outcome of cytogenetic tests. The latest version of ISCN was published in 2009. This implementation guide recommends using ISCN 2009 to represent cytogenetic test results; it allows previous versions such as ISCN 2005. Shaffer LG, Slovak ML, Campbell LJ, eds. ISCN 2009: An International System for Human Cytogenetic Nomenclature (2009): Recommendations of the International Standing Committee on Human Cytogenetic Nomenclature. Karger in collaboration with Cytogenetics and Genome Research; 2009.

6.1.1 Human Reference Sequence Assembly Release Number (required)

The human reference sequence assembly is the baseline sequence used by cytogenomic microarray analysis to report copy number variations. Genome Reference Consortium (GRC) determines the

releases of the human reference sequence assembly.⁵ The current human assembly release number is GRCh37. Previous releases include NCBI36, NCBI35.

⁵ <http://www.ncbi.nlm.nih.gov/projects/genome/assembly/grc/data.shtml>

7. Logical Message Types

7.1 INTRODUCTION AND STRATEGY

The design of the Cytogenetic Test Result Reporting message is consistent with the design of the Genetic Test Result Reporting message as specified in the *HL7 Version 2 implementation Guide: Clinical Genomics; Fully LOINC-Qualified Genetic Variation Model, Release 1*. A similar LOINC panel approach has been applied to the cytogenetic test result message structure. The Cytogenetic Test Result Reporting message is defined by a set of LOINC panels, which serve as templates for the messages. In general, LOINC panel definitions include one LOINC code to identify the whole panel, and a set of LOINC codes for each child element of that panel. A child element can also be a LOINC panel, and such panels can repeat to provide a structure that can accommodate many reporting patterns. For each such child element, the panel definition also includes its data type, optionality, and answer list. The definitional information for the four panels used to report cytogenetics test result reports is included in this guide. It can also be obtained in electronic form from the LOINC web site. In a message, each new panel of observations begins with an OBR segment that carries the LOINC ID for that panel. It is followed by a series of OBX's each of which carries the LOINC ID (OBX-3), and the value (OBX-5) of a particular observation. Nested OBR segments are used to reflect the LOINC panel structures. To create the nesting, OBR-29 is used to link the current OBR to its parent OBR's filler order number.

The cytogenetic LOINC panel structure starts with the *chromosome analysis master panel*. The master panel contains a required *chromosome analysis summary panel*, and at least one of the following: the *chromosome analysis G-banding panel*, the *chromosome analysis FISH panel*, or the *chromosome analysis arr copy number change panel*. The master panel allows the laboratory to report results of individual G-banding, FISH, or an arr copy number change test by itself, or as two or three tests combined. For the first OBR segment in a message, OBR-4 is the order code. The LOINC code 62389-2 *Chromosome Master Panel* is transmitted in the OBR-50 parent *universal service identifier* field.

The *chromosome analysis summary panel* will always be used to report the overall summary of the test. If only one method (G-banding, FISH, or arr) is used during the chromosome analysis, the optional *chromosome analysis summary panel* that is contained under each G-banding, FISH, or arr copy number change panel should NOT be used. For any test, if multiple testing methods are used, then the *chromosome analysis summary panel* at the higher level would allow an overall summary to be reported, and the chromosome analysis summary panel at the individual test level will allow summary for the individual tests to be reported.

In addition to the summary panel, G-banding, FISH, and arr copy number change panels include discrete information that is specific to its technique. For example, it is important to report the *human reference sequence release number* for an arr analysis. This indicates which version of the human assembly is used. The human reference release number released by the GRC is the standard for human reference sequence.

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

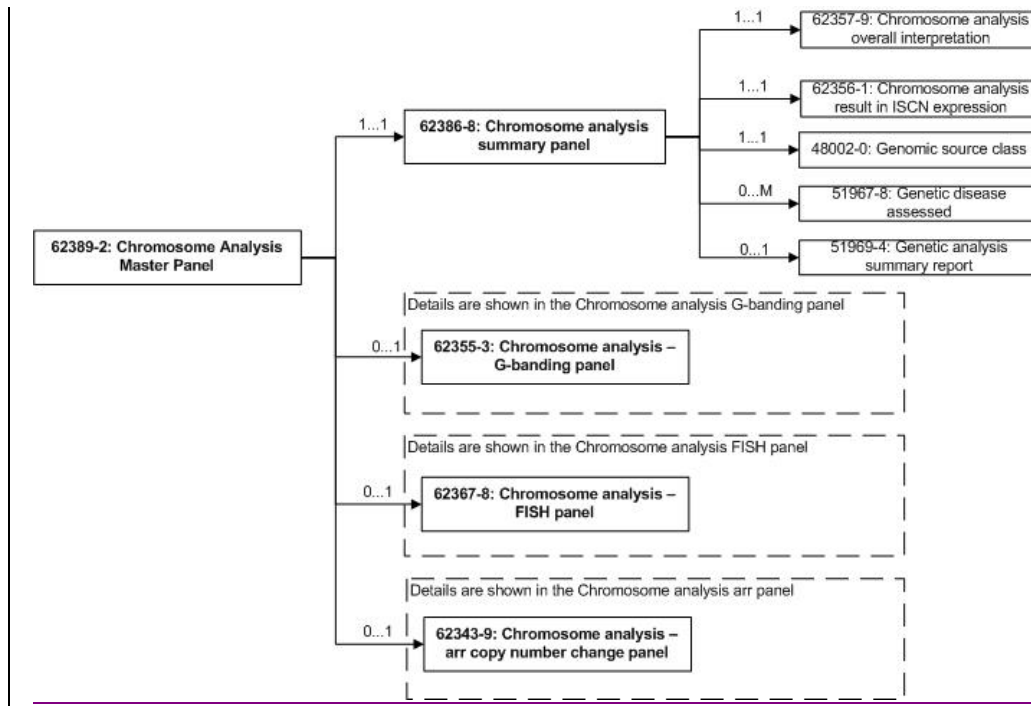


Figure 1: Chromosome analysis master panel

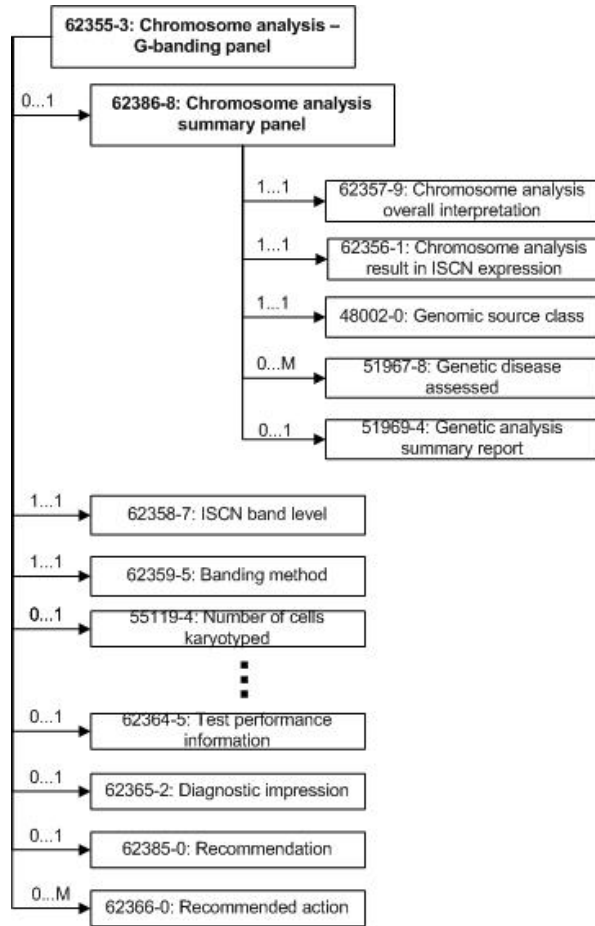


Figure 2: Chromosome analysis G-banding panel

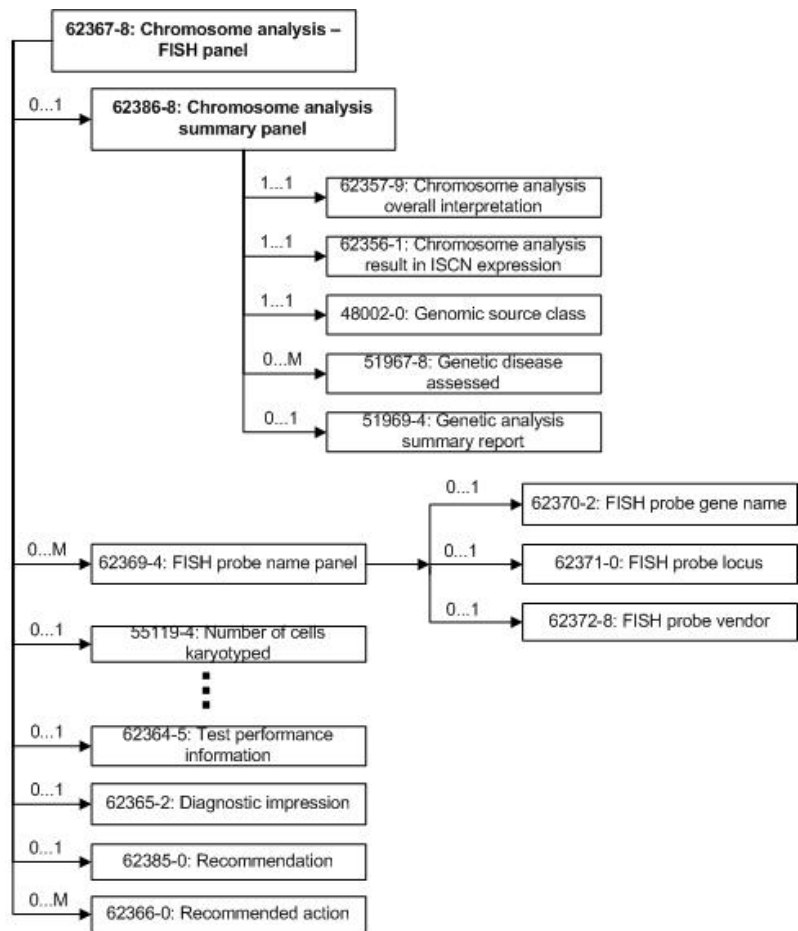


Figure 3: Chromosome analysis FISH panel

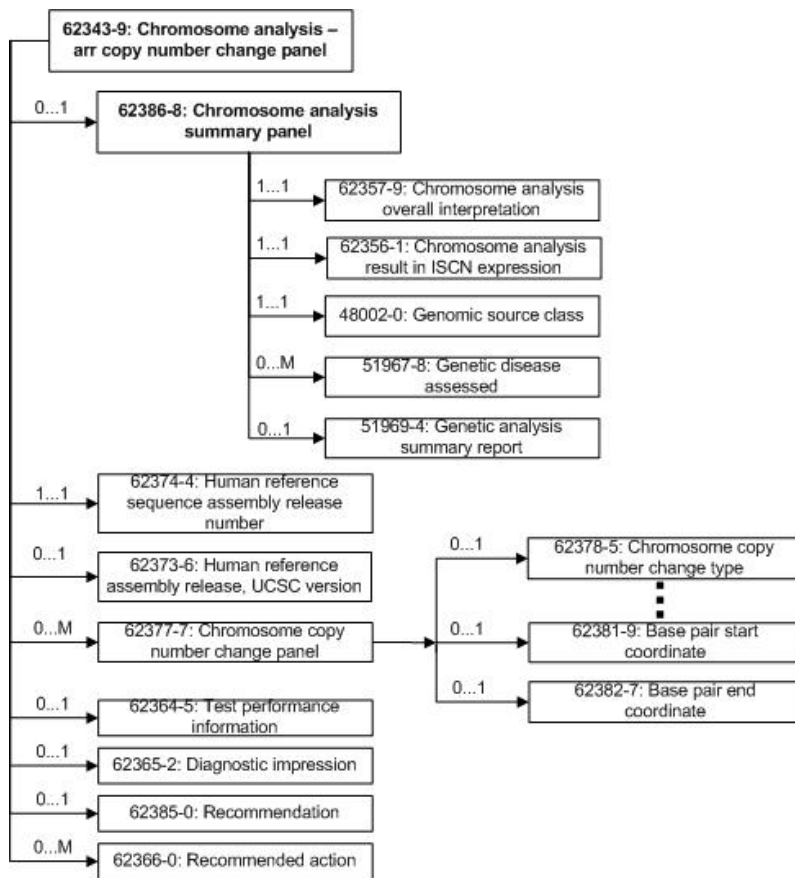


Figure 4: Chromosome analysis arr copy number change panel

7.2 MESSAGE DEFINITIONS

The complexity of cytogenetic data requires additional coding of the message components using LOINC. These codes are listed in tables 7.1 to 7.7. As stated in the Introduction, LOINC coding has several advantages, including more robust representation of the data when persisted in a database, increased accuracy when supporting multiple HL7 message formats, consistency for clinical decision support, and applicability to many complex reporting requirements.

7.3 MESSAGE COMPONENTS

PLEASE NOTE:

The following tables 7-1 through 7-7 are NOT segment definitions. They specify the content of LOINC panels.

7.3.1 Test Interpretation

You can view the list of the individual test components included in each panel. The elements will be accompanied by a usage flag that will denote the expected appearance of the panel element in the panel when result. A usage flag is always one of three states:

- **Required.** The panel element is always expected to be reported when the panel is result.
- **Optional.** Report if available. The panel element may not be reported with a panel result depending upon institutional policy or capabilities of the reporting lab.
- **Conditional.** The panel element is a key finding in the panel report and should be assumed to be negative, absent or not present if the panel result does not include data for this element.

7.3.1.0 Chromosome Analysis Master Panel

The *Chromosome Analysis Master Panel* is an OBR which must contain a child OBR with chromosome analysis summary panel, and optional child OBR(s) with either chromosome analysis G-banding test results, chromosome analysis FISH test results, and/or chromosome analysis arr copy number change test results.

TABLE 7-1 – CHROMOSOME ANALYSIS MASTER PANEL							
OBR/OBX	DT	Usage	Cardinality	Value Set	LOINC Code	LOINC Element Name	Description/Comments
OBR	N/A	R	1	N/A	62389-2	Chromosome Analysis Master Panel	This is the parent OBR for the panel holding the summary of chromosome analysis (i.e. Chromosome Analysis Summary Panel).

7.3.1.1 Chromosome Analysis Summary Panel

The Chromosome Analysis Summary Panel is an OBR which must contain the following required OBXs: chromosome analysis result in ISCN expression, chromosome analysis overall interpretation, genomic source class, and genetic analysis summary report.

TABLE 7-2 – CHROMOSOME ANALYSIS SUMMARY PANEL							
OBR/OBX	DT	Usage	Cardinality	Value Set	LOINC Code	LOINC Element Name	Description/Comments
OBR	N/A	R	1	N/A	62386-8	Chromosome analysis summary panel	The summary panel for a chromosome analysis for one or more laboratory tests (e.g. G-banded, FISH, or arr copy number change) on a single accession.
OBX	CWE	R	1	N/A	62356-1	Chromosome analysis result in ISCN expression	The chromosome analysis result, which is expressed using the International System for Human Cytogenetics Nomenclature (ISCN).

OBX	CWE	R	1	LOINC	62357-9	Chromosome analysis overall interpretation	The overall interpretation of a chromosome analysis
OBX	CWE	R	1	LOINC	48002-0	Genomic source class	The genomic class of the specimen being analyzed: germline for inherited genome, somatic for cancer genome, and prenatal for fetal genome.
OBX	CWE	O	0-M	SNOMED	51967-8	Genetic disease assessed	A coded disease (recommend SNOMED) which is known to be caused by or identified by genomic DNA Markers
OBX	FT	R	1	N/A	51969-4	Genetic analysis summary report	Narrative report in disease diagnostic-based format.

7.3.2 Findings

7.3.2.0 Chromosome Analysis G-banded panel

The chromosome analysis G-banded panel reports findings that are specific to conventional G-banding chromosome analysis, such as ISCN band level. *ISCN band level* is the required information for correct interpretation of the results.

TABLE 7-3 – CHROMOSOME ANALYSIS G-BANDED PANEL

OBR/OBX	DT	Usage	Cardinality	Value Set	LOINC Code	LOINC Element Name	Description/Comments
OBR	N/A	R	1	N/A	62355-3	Chromosome analysis G-banding panel	Chromosome analysis that uses traditional banding technique.
OBX	CWE	R	1	LOINC	62358-7	ISCN band level	The ISCN band level indicates the banding resolution that was used to interpret the karyotyping results. Example levels are 450, 800. The higher the number, the higher the resolution.
OBX	CWE	R	1	LOINC	62359-5	Banding method	The banding method that is used to karyotype.
OBX	NM	O	0-1	N/A	55199-4	Number of cells karyotyped	Number of cells that were karyotyped when the test was performed.

OBX	NM	O	0-1	N/A	62360-3	Number of cells analyzed	Number of analyzed cells. Analyzed cells are banded metaphase cells in which the individual chromosomes are counted and evaluated in their entirety, either at the microscope or from intact digitized images or photographic prints.
OBX	NM	O	0-1	N/A	62361-1	Number of cells counted	The number of metaphase cells evaluated for chromosome number.
OBX	NM	O	0-1	N/A	62362-9	Number of colonies counted	Number of colonies that were counted. Colony is a discrete focus of cells that is harvested and stained while attached to the cell culture growth substrate.
OBX	CWE	O	0-1	LOINC	62363-7	Mosaicism detected	This is a Yes and No indicator to specify that mosaicism was detected.
OBX	FT	O	0-1	N/A	62364-5	Test performance information	The narrative description about the test. This is a report section header.
OBX	FT	O	0-1	N/A	62365-2	Diagnostic impression	The narrative description about the diagnostic impression. This is a report section header.
OBX	FT	O	0-1	N/A	62385-0	Recommendation	The recommendation of a test or study. This is a report section header.
OBX	CWE	O	0-M	N/A	62366-0	Recommended action	A coded list of recommended actions based on the test/study result.

7.3.2.1 Chromosome Analysis FISH panel

The chromosome analysis FISH panel reports findings that are specific to chromosome analysis that use the FISH technique, such as FISH probe name panel.

TABLE 7-4 – CHROMOSOME ANALYSIS FISH PANEL							
OBR/ OBX	OBX-2 Value Type	Usage*	Cardinality	Value Set	LOINC Code	LOINC Element Name	Description/Comments
OBR	N/A	R	1	N/A	62367-8	Chromosome analysis FISH panel	The constitutional chromosome analysis, which uses FISH technique.

OBX	CWE	O	0-1	LOINC	62368-6	Cell phase	
OBR	N/A	O	0-M	N/A	62369-4	FISH probe name panel	The panel code that contains the FISH probe gene name, FISH probe locus, or FISH probe vendor.
OBX	NM	O	0-1	N/A	62360-3	Number of cells analyzed	Number of analyzed cell.
OBX	FT	O	0-1	N/A	62364-5	Test performance information	The narrative description about the test. This is a report section header.
OBX	FT	O	0-1	N/A	62365-2	Diagnostic impression	The narrative description about the diagnostic impression. This is a report section header.
OBX	FT	O	0-1	N/A	62385-0	Recommendation	The recommendation of a test or study. This is a report section header.
OBX	CWE	O	0-M	N/A	62366-0	Recommended action	A coded list of recommended actions based on the test/study result.

7.3.2.2 Chromosome Analysis arr Copy Number Change panel

The *chromosome analysis arr copy number change panel* reports findings that are specific to arr, such as human reference sequence assembly release number. *ISCN band level* is the required information for correct interpretation of the results.

TABLE 7-5 – CHROMOSOME ANALYSIS ARR COPY NUMBER CHANGE PANEL							
OBR/OBX	OBX-2 Value Type	Usage*	Cardinality	Value Set	LOINC Code	LOINC Element Name	Description/Comments
OBR	N/A	R	1	N/A	62343-9	Chromosome analysis arr panel	The microarray chromosome analysis for copy number change.
OBX	CWE	R	1	LOINC	62374-4	Human reference sequence assembly release number	The release number of the human sequence assemblies. The current list includes: GRCh37, NCBI Build 36.1, NCBI Build 35, NCBI Build 34
OBX	CWE	O	0-1	LOINC	62373-6	Human reference sequence assembly release, UCSC version	The human genome assembly release number. This is the release number published by University of California Santa Cruz (UCSC). The UCSC human genome

							assemblies with release number hg10 and above are identical to the NCBI assemblies. Refer to http://genome.ucsc.edu/FAQ/FAQreleases.html for the correspondence between UCSC human genome assembly release number and NCBI human genome assembly build numbers. The list of UCSC release numbers are: hg19, hg18, hg17, hg16
OBX	FT	O	0-1	N/A	62375-1	Microarray platform	The name of the microarray platform used for analysis.
OBX	FT	O	0-1	N/A	62376-9	Microarray platform version number	The version number of the microarray platform.
OBR	N/A	O	0-M	N/A	62377-7	Chromosome copy number change panel	The panel code that is used to contain the details about the copy number change, such as the base pair start and end coordinates of where the change occurred.
OBX	FT	O	0-1	N/A	62364-5	Test performance information	The narrative description about the test. This is a report section header.
OBX	FT	O	0-1	N/A	62365-2	Diagnostic impression	The narrative description about the diagnostic impression. This is a report section header.
OBX	FT	O	0-1	N/A	62385-0	Recommendation	The recommendation of a test or study. This is a report section header.
	CWE	O	0-M	N/A	62366-0	Recommended action	A coded list of recommended actions based on the test/study result.

7.3.2.3 FISH probe name panel

The FISH probe name panel has a cardinality of 0-M (zero to many). If multiple FISH probes are used for the analysis, multiple instances of FISH probe name panels will be reported.

TABLE 7-6 – FISH PROBE NAME PANEL							
OBR/ OBX	OBX-2 Value Type	Usage*	Cardinality	Value Set	LOINC Code	LOINC Element Name	Description/Comments
OBR	N/A	R	0-M	N/A	62369-4	FISH probe name panel	A LOINC panel code that is used to contain FISH probe related data elements.
OBX	CWE	O	0-1	N/A	62370-2	FISH probe gene name	The FISH probe targeted gene.
OBX	CWE	O	0-1	N/A	62371-0	FISH probe locus name	The FISH probe targeted locus.
OBX	CWE	O	0-1	N/A	62372-8	FISH probe vendor	The vendor who supplied the FISH probe.

7.3.2.4 Chromosome Copy Number Change panel

Chromosome copy number change panel reports detailed information about a specific chromosome copy number change. It is required to report the chromosome copy number change type, whether the change is chromosome gain or loss.

TABLE 7-7 – CHROMOSOME ANALYSIS ARR COPY NUMBER CHANGE PANEL							
OBR/ OBX	OBX-2 Value Type	Usage*	Cardinality	Value Set	LOINC Code	LOINC Element Name	Description/Comments
OBR	N/A	R	0-M	N/A	62377-7	Chromosome copy number change panel	The panel code that is used to contain details about the copy number change, such as the base pair start and end coordinates where the change occurred.
OBX	CWE	R	1	LOINC	62378-5	Chromosome copy number change type	Gain, Loss (Duplication, Deletion)
OBX	CWE	O	0-1	N/A	62379-3	Chromosome band involved start	The start point of the chromosome band that is involved in the copy number change.
OBX	CWE	O	0-1	N/A	62380-1	Chromosome band involved end	The end point of the chromosome band that is involved in the copy

							number change.
OBX	NM	O	0-1	N/A	62381-9	Base pair start coordinate	The start coordinate of the base pair.
OBX	NM	O	0-1	N/A	62382-7	Base pair end coordinate	The end coordinate of the base pair.
OBX	CWE	O	0-1	N/A	62383-5	Flanking normal region before start	The normal DNA sequences extending before the start of a specific chromosome copy number change.
OBX	CWE	O	0-1	N/A	62384-3	Flanking normal region after end	The normal DNA sequences extending after the end of a specific chromosome copy number change.

7.4 LOINC CODES

TABLE 7-8 – LOINC CODES

LOINC #	Component	Property	Time	System	Scale	Method
62389-2	Chromosome analysis master panel	-	Pt	Bld/Tiss	-	Molgen
62386-8	Chromosome analysis summary panel	-	Pt	Bld/Tiss	-	Molgen
62356-1	Chromosome analysis result in ISCN expression	Find	Pt	Bld/Tiss	Nom	Molgen
62357-9	Chromosome analysis overall interpretation	Imp	Pt	Bld/Tiss	Nom	Molgen
48002-0	Genomic source class	Type	Pt	Bld/Tiss	Nom	Molgen
51967-8	Genetic disease assessed	ID	Pt	Bld/Tiss	Nom	Molgen
51969-4	Genetic analysis summary report	Find	Pt	Bld/Tiss	Doc	Molgen
62355-3	Chromosome analysis	-	Pt	Bld/Tiss	-	Molgen. banding
62358-7	ISCN band level	Find	Pt	Bld/Tiss	Ord	Molgen
62359-5	Banding method	Type	Pt	Bld/Tiss	Nom	Molgen
62360-3	Number of cells analyzed	Num	Pt	Bld/Tiss	Qn	Molgen
62361-1	Number of cells counted	Num	Pt	Bld/Tiss	Qn	Molgen
55199-4	Number of cells karyotyped	Num	Pt	Bld/Tiss	Qn	Molgen
62362-9	Number of colonies counted	Num	Pt	Bld/Tiss	Qn	Molgen
62363-7	Mosaicism detected	Find	Pt	Bld/Tiss	Nom	Molgen
62364-5	Test performance information	Find	Pt	Bld/Tiss	Xxx	Doc
62365-2	Diagnostic impression	Imp	Pt	Bld/Tiss	Xxx	Doc
62385-0	Recommendation	Imp	Pt	Bld/Tiss	Xxx	Doc

TABLE 7-8 – LOINC CODES						
LOINC #	Component	Property	Time	System	Scale	Method
62366-0	Recommended action	Find	Pt	Bld/Tiss	Xxx	Doc
62367-8	Chromosome analysis	-	Pt	Bld/Tiss	-	Molgen. FISH
62368-6	Cell phase	Type	Pt	Bld/Tiss	Nom	Molgen
62369-4	FISH probe name panel	-	Pt	Bld/Tiss	-	Molgen
62370-2	FISH probe gene name	ID	Pt	Bld/Tiss	Nom	Molgen
62371-0	FISH probe locus	ID	Pt	Bld/Tiss	Nom	Molgen
62372-8	FISH probe vendor	ID	Pt	Bld/Tiss	Nom	Molgen
62343-9	Chromosome analysis microarray copy number change	-	Pt	Bld/Tiss	-	Molgen. arr
62373-6	Human reference sequence assembly release, UCSC version	Id	Pt	Bld/Tiss	Nom	Molgen
62374-4	Human reference sequence assembly release number	Id	Pt	Bld/Tiss	Nom	Molgen
62375-1	Microarray platform	Id	Pt	Bld/Tiss	Nar	Molgen
62376-9	Microarray platform version number	Id	Pt	Bld/Tiss	Nom	Molgen
62377-7	Chromosome copy number change panel	-	Pt	Bld/Tiss	-	Molgen
62378-5	Chromosome copy number change type	Find	Pt	Bld/Tiss	Nom	Molgen
62379-3	Chromosome band involved start	Find	Pt	Bld/Tiss	Nom	Molgen
62380-1	Chromosome band involved end	Find	Pt	Bld/Tiss	Nom	Molgen
68381-9	Base pair start coordinate	Num	Pt	Bld/Tiss	Qn	Molgen
68382-7	Base pair end coordinate	Num	Pt	Bld/Tiss	Qn	Molgen
62383-5	Flanking normal region before start	Find	Pt	Bld/Tiss	Nom	Molgen
62384-3	Flanking normal region after end	Find	Pt	Bld/Tiss	Nom	Molgen

7.5 LOINC ANSWER LISTS

TABLE 7-9 – LOINC ANSWER LISTS				
LOINC code	LOINC component	Sequence	Answer text	LOINC answer code
62357-9	Chromosome analysis overall interpretation	1	Normal	LA6626-1
		2	Abnormal	LA12748-2
		3	Clinical significance unknown	LA14007-1
48002-0	Genomic source class	1	Germline	LA6683-2

TABLE 7-9 – LOINC ANSWER LISTS				
LOINC code	LOINC component	Sequence	Answer text	LOINC answer code
		2	Somatic	LA6684-0
		3	Prenatal	LA6685-7
62358-7	ISCN band level	1	400	LA14008-9
		2	500	LA14009-7
		3	600	LA14010-5
		4	800	LA14011-3
		5	850	LA14012-1
		6	425	LA14112-9
		7	450	LA14113-7
		8	550	LA14114-5
		9	575	LA14115--2
		10	650	LA14116-0
62359-5	Banding method	1	G-banding	LA14013-9
		2	R-banding	LA14015-7
		3	Q-banding	LA14015-4
		4	C-banding	LA14016-2
		5	T-banding	LA14017-0
62366-0	Recommended action	1	Genetic counseling recommended	LA14020-4
		2	Confirmatory testing recommended	LA14021-2
		3	Additional testing recommended	LA14022-0

7.6 SPECIAL SYNTAX

An International System for Human Cytogenetic Nomenclature (ISCN)

The International System for Human Cytogenetic Nomenclature (ISCN) was created by the International Standing committee on Human Cytogenetic Nomenclature to represent the results of cytogenetic tests. The latest version of ISCN was published in 2009. One of the aims of ISCN is to prevent confusion in reporting research cytogenetics results. ISCN is accepted as a standard within the industry. It specifies the nomenclature to describe karyotypes, chromosome abnormalities, in situ hybridization, etc.

Examples of ISCN notations:

46,XY

Normal male.

45,XX,-22

Female monosomy 22.

ish del(22)(q13.3q13.3)(ARSA-)

A deletion of distal 22q was identified by in situ Hybridization (ish) using a probe to the ARSA locus.

arr 17p11.2(16,512,256-20,405,113)x3 dn

Microarray analysis shows a single copy gain of the short arm of chromosome 17 at band p11.2. The duplication is ~3.9 Mb in size and is de novo in origin.

8. Example Genetic Test Laboratory Messages

Note: This chapter is based on Chapter 7 (Example Laboratory Result Messages) of the parent implementation guide entitled 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. The guide can be found at http://www.hl7.org/Memonly/downloads/Standards_Messaging_V251/InteroperabilitySpecificationLabResultMessage_v251.zip (HL7 membership required).

The examples in this section adhere to HL7 publishing requirements in that all persons and facilities are fictitious. They are taken from the *HL7 Version 3 Publishing Facilitator's Guide, Appendix D, Storyboard Names*.

Emphasis has also been placed on demonstrating the use of standardized vocabularies together with local terminology.

8.1 MINIMAL MESSAGE WITH ACKNOWLEDGEMENT

For Information on 'Minimal Message with Acknowledgement' see Chapter 7 (Example Laboratory Result Messages) of the parent implementation guide entitled 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. The guide can be found at http://www.hl7.org/Memonly/downloads/Standards_Messaging_V251/InteroperabilitySpecificationLabResultMessage_v251.zip (HL7 membership required). This includes Successful Receipt Message, Error on Receipt Message, and Reject Receipt Message.

8.2 CHROMOSOME ANALYSIS G-BANDING TEST RESULT MESSAGE

8.2.1 Example: Trisomy 21 (Down syndrome), Male

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.

PID-->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||PO-1000^ARUP|200291^Chromosome analysis chorionic villus sampling^99ARU-ORDER-TEST-ID||20100702000000|20100702100909|||||201070201410||12345^Dr.Jones|||||20080703000000||F|||||^Fetal demise|||||||||||||62389-2^Chromosome analysis master panel^LN

SPM|1|||^Placental tissue - Villi|||||||||20100702100909

OBR|2||PO-1000-1^ARUP|62355-3^Chromosome analysis G-banding^LN||20100702000000|20100702100909|||||201070201410||12345^Dr.Jones|||||201070201410||F||||PO-1000^ARUP

OBX|1|CWE|62358-7^SCN band level^LN||LA14112-9^425^LN|||||F|201070201410|||||||ARUP Laboratories

OBX|2|CWE|62359-5^Banding method^LN||LA14013-9^G-banding^LN||||F|20080702100909|||||
|||ARUP Laboratories

OBX|3|NM|62361-1^Numer of cells counted^LN||20||||F|201070201410|||||||ARUP Laboratories

OBX|4|CWE|62366-0^Recommended action^LN||LA14020-4^Genetic counseling recommended^LN|
||||F|201070201410|||||||ARUP Laboratories

OBX|5|FT|62385-0^Recommendation^LN||1. Genetic counseling. 2. Monitor subsequent pregnancies
with prenatal diagnosis||||F|201070201410|||||||ARUP Laboratories

(... more OBXs could be placed here to represent other information in the G-banding panel...)

OBR|3||PO-1000-2^ARUP|62386-8^Chromosome analysis summary panel^LN||20100702000000
|20100702100909|||||201070201410||12345^Dr.Jones|||||201070201410||F||||PO-1000^ARUP

OBX|1|CWE|62357-9^Chromosome analysis result overall interpretation^LN||LA6626-
1^Normal^LN||||F|201070201410|||||||ARUP Laboratories

OBX|2|CWE|62356-1^Chromosome analysis result in ISCN
expression^LN||47,XY,+21^2.16.840.1.113883.6.299^^2005||||F|201070201410|||||||ARUP
Laboratories

OBX|3|CWE|48002-0^Genomic source class^LN||LA6683-3^Prenatal^LN||||F|201070201410
|||||||ARUP Laboratories

8.3 CHROMOSOME ANALYSIS FISH TEST RESULT MESSAGE

8.3.1 Example: 22q.11.2 Deletion Syndrome (DiGeorge syndrome)

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.

PID-->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||PO-2000^ARUP|2002299^Chromosome analysis, FISH metaphase^ARUP^99ARU-ORDER-
TEST-ID||20100702000000|20100702100909|||||201070201410||12345^Dr.Jones|||||
20080703000000||F||||||^Other developmental speech|||||||62389-2^Chromosome analysis
master panel^LN

SPM|1||^Peripheral blood|||||||20100702100909

OBR|2||PO-2000-1^ARUP|62367-8^Chromosome analysis FISH panel^LN||20100702000000
|20100702100909|||||201070201410||12345^Dr.Jones|||||201070201410||F||||PO-2000^ARUP

OBX|1|CWE|62366-0^Recommended action^LN||LA14020-4^Genetic counseling recommended^LN|
||||F|201070201410|||||||ARUP Laboratories

(... more OBXs could be placed here to represent other information in the FISH panel...)

OBR|3||PO-2000-2^ARUP|62386-8^Chromosome analysis summary panel^LN||20100702000000
|20100702100909|||||201070201410||12345^Dr.Jones|||||201070201410||F||||PO-2000^ARUP

OBX|1|CWE|62357-9^Chromosome analysis result overall interpretation^LN
||LA_X4^Abnormal^LN||||F|201070201410|||||||ARUP Laboratories

OBX|2|CWE|62356-1^Chromosome analysis result in ISCN expression^LN||ish
del(22)(q11.2q11.2)(H1RA-)^ISCN-2005||||F|201070201410|||||||ARUP Laboratories

OBX|3|CWE|48002-0^Genomic source class^LN||LA6683-2^Germline^LN||||F|201070201410
|||||||ARUP Laboratories

(... more OBXs could be placed here to represent other information in the summary panel...)

8.4 CHROMOSOME ANALYSIS ARR COPY NUMBER CHANGE TEST RESULT MESSAGE

8.4.1 Example:

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.

PID-->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||PO-1001^ARUP|0040201^Genomic Microarray, U-Array Chip^99ARU-ORDER-TEST-ID||20100702000000
|20100702100909|||||201070201410||12345^Dr.Jones|||||20080703000000||F|||||^Other developmental speech|||||||||||||62389-2^Chromosome analysis master panel^LN |

SPM|1|||^Peripheral blood||||||||||20100702100909

OBR|2||PO-1001-1^ARUP|62377-7^Chromosome analysis arr copy number change panel^LN
||20100702000000|20100702100909|||||201070201410||12345^Dr.Jones|||||201070201410||F||||
PO-1001^ARUP

OBX|1|CWE|62374-4^Human reference sequence NCBI build id^LN||LA_X5^NCBI35^LN|||||F|
201070201410|||||||||ARUP Laboratories

OBX|2|CWE|62375-1^Arr platform^LN||^U-Array Cyto6000|||||F|201070201410|||||||||ARUP
Laboratories

(... more OBXs could be placed here to represent other information in the arr panel...)

OBR|3||PO-1001-2^ARUP|62386-8^Chromosome analysis summary panel^LN||20100702000000
|20100702100909|||||201070201410||12345^Dr.Jones|||||201070201410||F||||PO-1001^ARUP

OBX|1|CWE|62357-9^Chromosome analysis result overall interpretation^LN
||LA_X4^Abnormal^LN|||||F|201070201410|||||||||ARUP Laboratories

OBX|2|CWE|62356-1^Chromosome analysis result in ISCN expression^LN||arr cgh
1q21.1(143,612,538bp->145,024,147bp)x1^2.16.840.1.113883.6.299^^^2005|||||F|201070201410
|||||||||ARUP Laboratories

OBX|3|CWE|48002-0^Genomic source class^LN||LA6683-2^Germline^LN|||||F|201070201410
|||||||||ARUP Laboratories

OBR|4||PO-1001-3^ARUP|62377-7^Chromosome copy number change panel^LN||20100702000000
|20100702100909|||||201070201410||12345^Dr.Jones|||||201070201410||F||||PO-1001-2^ARUP

OBX|1|CWE|62378-5^Chromosome analysis copy number change type^LN||LA_X7^Deletion^LN|
|||||F|201070201410|||||||||ARUP Laboratories

(... more OBXs could be placed here to represent other information in the copy number change panel...)

9.Future Plans

This implementation guide has focused on constitutional cytogenetic test result reporting. Future releases of this implementation guide will be released in order to extend the HL7 2.5.1 cytogenetic test reporting functionality to support reporting of cancer cytogenetic test results and SNP array.

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