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

Draft Standard for Trial Use
Second Ballot
May 2011
CDAR2_IG_GENTESTRPT_R1_O2_2011MAY
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## Authors

<table>
<thead>
<tr>
<th>Role</th>
<th>Name</th>
<th>Affiliation</th>
<th>Email</th>
</tr>
</thead>
<tbody>
<tr>
<td>Primary Editor</td>
<td>Amnon Shabo, Ph.D.</td>
<td>IBM Research Lab in Haifa</td>
<td><a href="mailto:SHABO@il.ibm.com">SHABO@il.ibm.com</a></td>
</tr>
<tr>
<td>Co-Editor</td>
<td>Mollie Ullman-Cullere MS, MSE</td>
<td>Department of Clinical Research Informatics</td>
<td><a href="mailto:Mollie_Ullman-Cullere@dfci.harvard.edu">Mollie_Ullman-Cullere@dfci.harvard.edu</a></td>
</tr>
<tr>
<td>CG and SDWG Members</td>
<td></td>
<td>If you're interested in contributing to the development of the GTR IG, please contact Amnon Shabo.</td>
<td></td>
</tr>
</tbody>
</table>

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Acknowledgments

We acknowledge the foundational work on HL7 Version 3 and the Reference Information Model (RIM), the HL7 domain committees, especially the work done by the Structured Documents Work Group on Clinical Document Architecture (CDA) itself.

We acknowledge the efforts of the HL7 Clinical Genomics Work Group which has been developing v3 specifications for the past seven years as well as v2 implementation guide for genetic testing results message. Note that the LOINC codes developed within the v2 effort are utilized in this GTR IG.

We acknowledge the efforts the MDHT tool developers who work closely and tirelessly with us to accommodate the requirements of the GTR.

LOINC® is a registered United States trademark of Regenstrief Institute, Inc.
<table>
<thead>
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<th>Rev</th>
<th>Date</th>
<th>By Whom</th>
<th>Changes</th>
</tr>
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<td>Amnon Shabo</td>
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<td>Amnon Shabo</td>
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</tr>
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<td>CGWG review</td>
</tr>
<tr>
<td>Draft 3</td>
<td>March 2011</td>
<td>Amnon Shabo</td>
<td>CGWG review</td>
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The purpose of this Implementation Guide (IG) is to specify a standard for Genetic Testing Reports.

In this project, the Clinical Genomics and Structured Documents Work Groups will jointly develop a CDA Implementation Guide (IG) for genetic testing reports.
**Purpose**

Genetic tests have recently become an important tool in clinical care that further personalizes the care processes based on the patient individual genetic makeup. Genetic testing methods are diverse and span from testing for known germline mutations in the context of single-gene disorders, to full sequencing of genes in tumor tissues looking for somatic variations in cancer cells. We also see the emerging use of gene expression testing in clinical care and it is expected to see a growing use of research techniques adjusted to healthcare.

As a consequence of that diversity and the constantly growing number of techniques yielding new result formats less familiar to the receiving party, we see existing report formats having emphasis on detailed but easy-to-understand interpretations of the testing results along with concrete recommendations. They also provide, within the report itself, detailed information on the tests performed including references to the appropriate scientific studies and publications in a format that looks quite often like a short abstract in a scientific journal.

Within the clinical environment, genetic test results typically flow from the genetic testing laboratory into the electronic health record (EHR). From the EHR these results may flow into another EHR or a personal health record (PHR). In some realms the first transmission of this data (from the lab into the EHR) is performed using the Laboratory 2.5.1 message standard. Clinical Genomics has written an implementation guide which extends this standard for the support of clinical genetics (HL7 Version 2 Implementation Guide: Clinical Genomics; Fully LOINC-Qualified Genetic Variation Model, Release 1 (US Realm)).

In some realms, the second transmission of this data (EHR to EHR/PHR) is performed using the CCD message model (a constrained version of the CDA model). As such for the healthcare specific message, this implementation guide will minimally detail how certain data sets defined in the above mentioned implementation guide would be included using the CDA model as appropriate to the level of granularity of this human-readable report.

Note: The producers of GTR documents include genetic labs as well as clinical geneticists or any clinician who needs to create a report summarizing genetic testing results (and is capable and authorized to do so). In addition, all roles in a research environment that needs to summarize genetic assays are included in the scope.

**Approach**

The following GTR IG design principles are based on requirements analysis through collecting requirements from various stake-holders of genetic testing reports.

- Emphasize interpretations and recommendations (in the clinical environment, interpretations should be related to clinically relevant findings)
- Provide inline information on tests performed
- Represent interpretation by utilizing patterns of genotype-phenotype associations in the HL7 v3 Clinical Genomics and implement them as harmonized clinical statement entry-level templates in this IG
- Reference HL7 Clinical Genomics instances as the place holders of raw data (personal evidences) similarly to referencing images

In particular, the GTR is organized as follows:

- The rendered portion of the GTR (aka "narrative") is placed in the text attributes of sections and sub-sections
- The structured portion of the GTR is carried by the "ClinicalGenomicStatement" template (an abstract class). At its core, it has a genomic observation, optionally associated on the one side with indications for performing this observation and on the other side with interpretations of that observation
- Interpretations of a genomic observation are placed in sub-templates of the InterpretivePhenotypeObservation (an abstract class), for example, InterpretivePhenotypeObservationGeneticVariation
- A number of ClinicalGenomicStatement's can be placed in the TestDetailsSection which serves as a blueprint for specialized sections such as the GeneticVariationsSection or the PharmacogenomicSection
- Those specialized sections constitute the main layout of the GTR along with a summary section and a 'catcher' section titled "Other Tests" that can consist of test results that couldn't not be placed in one of the specialized sections
- Sections that don't have a sub-template of "ClinicalGenomicStatement" or of "InterpretivePhenotypeObservation", merely carry narrative content
Notes:
1. Where templates have undocumented attributes, these are TBD in later versions, or for documentation of how base CDA attributes are used in GTR, but are not conformance rules.
2. For XML snippets of the various templates with example values, please refer to the complete XML sample enclosed in the ballot package because the template examples in the guide are more of skeletal nature.
3. Constraints on the value attribute have been described in free text similar to Object Constraining Language (OCL) statements. In later versions of this guide, all constraints will be represented in OCL and thus could be validated.

Scope
The scope of this project is to define a universal implementation guide that can accommodate the needs described above which could then be further refined to specific genetic testing reports, either realm specific or method-specific or any other set of restrictions. In addition, this IG will strive to serve both research and clinical environment as much as possible.

Audience
The audience for this document includes software developers and implementers who wish to enable information exchange of genetic testing reports that can be both human readable and machine-processable.

Organization of This Guide
The requirements as laid out in the body of this document are subject to change per the policy on implementation guides (see section 13.02" Draft Standard for Trial Use Documents" within the HL7 Governance and Operations Manual, http://www.hl7.org/documentcenter/public/membership/HL7_Governance_and_Operations_Manual.pdf).

Templates
Templates are organized by document (see Document Templates), by section (see Section Templates), and by clinical statements (see Clinical Statement Templates). Within a section, templates are arranged hierarchically, where a more specific template is nested under the more generic template that it conforms to. See Templates by Containment for a listing of the higher level templates by containment; the appendix Templates Used in This Guide includes a table of all of the templates Organized Hierarchically.

Vocabulary and Value Sets
Vocabularies recommended in this guide are from standard vocabularies. The LOINC codes developed within the v2 genetic testing results message are used in this IG as optional value sets. When further constraining this IG to the US Realm, it would be possible to mandate the use of these LOINC value sets. Of note, these LOINC codes have been successfully piloted within the clinical genetic laboratory and EHR.

In addition, these terms have been added to the NCI-t (U.S. National Cancer Institute’s Thesaurus, see: http://ncit.nci.nih.gov/)

Use of Templates
When valued in an instance, the template identifier (templateId) signals the imposition of a set of template-defined constraints. The value of this attribute provides a unique identifier for the templates in question.

Originator Responsibilities
An originator can apply a templateId to assert conformance with a particular template.
In the most general forms of CDA exchange, an originator need not apply a `templateId` for every template that an object in an instance document conforms to. This implementation guide asserts when `templateIds` are required for conformance.

**Recipient Responsibilities**

A recipient may reject an instance that does not contain a particular `templateId` (e.g., a recipient looking to receive only GTR documents can reject an instance without the appropriate `templateId`).

A recipient may process objects in an instance document that do not contain a `templateId` (e.g., a recipient can process entries that contain Observation acts within a Problems section, even if the entries do not have `templateIds`).

**Conventions Used in This Guide**

**Conformance Requirements**

Conformance statements are grouped and identified by the name of the template, along with the `templateId` and the context of the template (e.g., ClinicalDocument, section, observation), which specifies the element under constraint. If a template is a specialization of another template, its first constraint indicates the more general template. In all cases where a more specific template conforms to a more general template, asserting the more specific template also implies conformance to the more general template. An example is shown below.

**Template name**

`[<type of template>: templateId <XXXX.XX.XXX.XXX>]`

Description of the template will be here.....

1. Conforms to `<The template name> Template (templateId: XXXX<XX>XXX>YYY).`
2. SHALL contain `[1..1] @classCode = <AAA> <code display name> (CodeSystem: 123.456.789 <XXX> Class) STATIC (CONF:<number>).`
3. ........

**Figure 1: Template name and "conforms to" appearance**

The conformance verb keyword at the start of a constraint (SHALL, SHOULD, MAY, etc.) indicates business conformance, whereas the cardinality indicator (0..1, 1..1, 1..*, etc.) specifies the allowable occurrences within an instance. Thus, "MAY contain 0..1" and "SHOULD contain 0..1" both allow for a document to omit the particular component, but the latter is a stronger recommendation that the component be included if it is known.

The following cardinality indicators may be interpreted as follows:

- 0..1 as zero to one present
- 1..1 as one and only one present
- 2..2 as two must be present
- 1..* as one or more present
- 0..* as zero to many present

Value set bindings adhere to HL7 Vocabulary Working Group best practices, and include both a conformance verb (SHALL, SHOULD, MAY, etc.) and an indication of DYNAMIC vs. STATIC binding. The use of SHALL requires that the component be valued with a member from the cited value set; however, in every case any HL7 "null" value such as other (OTH) or unknown (UNK) may be used.

Each constraint is uniquely identified (e.g., "CONF:605") by an identifier placed at or near the end of the constraint. These identifiers are not sequential as they are based on the order of creation of the constraint.

1. SHALL contain `[1..1] component/structuredBody (CONF:4082).`
   a. This component/structuredBody SHOULD contain `[0..1] component (CONF:4130) such that it
      a. SHALL contain `[1..1] Reporting Parameters section (templateId:2.16.840.1.113883.10.20.17.2.1) (CONF:4131).`
b. This component/structuredBody SHALL contain [1..1] component (CONF:4132) such that it

a. SHALL contain [1..1] Patient data section - NCR (templateId:2.16.840.1.113883.10.20.17.2.5) (CONF:4133).

Figure 2: Template-based conformance statements example

1. The value for "Observation / @moodCode" in a problem observation SHALL be "EVN"
2.16.840.1.113883.5.1001 ActMood STATIC. (CONF: 814).
3. The value for "Observation / statusCode" in a problem observation SHALL be "completed"
2.16.840.1.113883.5.14 ActStatus STATIC. (CONF: 816).
4. A problem observation SHOULD contain exactly one Observation / effectiveTime, to indicate the biological
timing of condition (e.g. the time the condition started, the onset of the illness or symptom, the duration of a
t condition). (CONF: 817).

Figure 3: Conformance statements example (taken from the CCD IG)

Keywords

The keywords SHALL, SHALL NOT, SHOULD, SHOULD NOT, MAY, and NEED NOT in this document are to be
interpreted as described in the HL7 Version 3 Publishing Facilitator's Guide:

• SHALL: an absolute requirement
• SHALL NOT: an absolute prohibition against inclusion
• SHOULD/SHOULD NOT: valid reasons to include or ignore a particular item, but must be understood and
carefully weighed
• MAY/NEED NOT: truly optional; can be included or omitted as the author decides with no implications

XML Examples

XML samples appear in various figures in this document in a fixed-width font. Portions of the XML content may be
omitted from the content for brevity, marked by an ellipsis (…) as shown in the example below.

```xml
<ClinicalDocument xmlns='urn:h17-org:v3'>
  ...
</ClinicalDocument>
```

Figure 4: ClinicalDocument example

XPath expressions are used in the narrative and conformance requirements to identify elements because they are
familiar to many XML implementers.

Contents of the DSTU Ballot Package

Table 1: Contents of the DSTU Ballot Package

<table>
<thead>
<tr>
<th>Filename</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>CDAR2_IG_GENTESTRPT_R1_O2_2011MAY.pdf</td>
<td>This guide</td>
</tr>
<tr>
<td>CDA-GeneticTestingReport-GeneticVariation-Sample-v7.xml</td>
<td>A Genetic Testing Report sample with Genetic Variation Sections</td>
</tr>
<tr>
<td>cda.xsl</td>
<td>A generic stylesheet for displaying the content of the sample document in HTML</td>
</tr>
</tbody>
</table>
Chapter 2

DOCUMENT TEMPLATES

Topics:

- Genetic Testing Report

This section contains the document level constraints for CDA documents that are compliant with this implementation guide.
Genetic Testing Report

The Genetic Testing Report 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. Most of them share a common structure represented by the Test Details Section which serves as a blueprint for most of the test-oriented sections like genetic variation or gene expression sections.

1. SHALL conform to CDA Clinical Document
2. SHALL contain [1..1] code/@code = "51969-4" Genetic analysis summary report (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-1)
   • Current LOINC code might be changed to reflect the mixed nature of the GTR, i.e., having both narrative and structured data.
3. SHOULD contain [0..1] component, such that it
   a. contains GTR Summary Section (templateId: 2.16.840.1.113883.10.20.20.1.1) (CONF-GTR-2)
4. SHOULD contain [0..1] component, such that it
   a. contains GTR Specimen Section (templateId: 2.16.840.1.113883.10.20.20.1.7) (CONF-GTR-3)
5. MAY contain [0..*] component, such that it
   a. contains GTR Genetic Variations Section (templateId: 2.16.840.1.113883.10.20.20.1.2) (CONF-GTR-4)
6. MAY contain [0..*] component, such that it
   a. contains GTR Gene Expression Section (templateId: 2.16.840.1.113883.10.20.20.1.3) (CONF-GTR-5)
7. MAY contain [0..*] component, such that it
   a. contains GTR Cytogenetics Section (templateId: 2.16.840.1.113883.10.20.20.1.4) (CONF-GTR-6)
8. SHALL contain [1..1] title (CONF-GTR-7)
   • Default title is "Genetic Testing Report".
9. MAY contain [0..*] component, such that it
   a. contains GTR Other Testing Section (templateId: 2.16.840.1.113883.10.20.20.1.6)

<?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">
   <templateId root="2.16.840.1.113883.10.20.20" assigningAuthorityName="GTR Genetic Testing Report"/>
   <code code="51969-4" codeSystem="2.16.840.1.113883.6.1" codeSystemName="LOINC" displayName="Genetic analysis summary report"/>
   <title/>
   <component>
     <structuredBody>
       <component>
         <section>
           <templateId root="2.16.840.1.113883.10.20.20.1.1"
                      assigningAuthorityName="GTR Summary Section"/>
           <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
                 codeSystemName="LOINC" displayName="Genetic Testing Summary Section"/>
           <title>Summary</title>
         </section>
       </component>
     </structuredBody>
   </component>
</ClinicalDocument>
Figure 5: Genetic Testing Report example
CHAPTER 3

SECTION TEMPLATES

Topics:

- Background Section
- Cytogenetics Section
- Findings Section
- Follow Up Genetic Tests Section
- Follow Up Visits To Specialists Section
- Gene Expression Section
- Genetic Variations Section
- Indications Section
- Interpretation Section
- Methodology Section
- Other Testing Section
- Overall Interpretation Section
- Recommendations Section
- Recommended Actions Section
- References Section
- Specimen Section
- Summary Section
- Test Details Section
- Test Information Section
- Tests Performed Section
**Background Section**

[Section: templateId 2.16.840.1.113883.10.20.20.1.9.1]

The Background Section is a narrative-only section. It nests within the TestInformationSection and its text attribute consists of narrative describing background of the genetic test at stake.

1. SHALL conform to CDA Section

2. SHALL contain [1..1] code/@code = "TBD" Genetic Testing Background Section (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-18)

3. SHALL contain [1..1] title (CONF-GTR-19)

```xml
<?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">
  <component>
    <structuredBody>
      <component/>
      <section>
        <templateId root="2.16.840.1.113883.10.20.20.1.9.1"
assigningAuthorityName="GTR Background Section"/>
        <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
codeSystemName="LOINC" displayName="Genetic Testing Background Section"/>
        <title>Genetic Testing Background Section</title>
      </section>
    </structuredBody>
  </component>
</ClinicalDocument>
```

Figure 6: Background Section example

**Cytogenetics Section**

[Section: templateId 2.16.840.1.113883.10.20.20.1.4]

The Cytogenetics Section resides at the highest level of the Genetic Testing Report and consists of data related to cytogenetics testing such as FISH.

1. SHALL conform to GTR Test Details Section template (templateId: 2.16.840.1.113883.10.20.20.1.8)

2. SHALL contain [1..1] code/@code = "TBD" Cytogenetics Section (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-25)

3. SHALL contain [1..1] title = "Cytogenetics" (CONF-GTR-26)

4. SHOULD contain [0..*] entry, such that it
   a. has @typeCode="COMP" COMP (component)
   b. contains GTR Clinical Genomic Statement Cytogenetics (templateId: 2.16.840.1.113883.10.20.20.2.2)

```xml
<?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">
  <component>
    <structuredBody>
      <component/>
      <section>
        <templateId root="2.16.840.1.113883.10.20.20.1.8"
assigningAuthorityName="GTR Test Details Section"/>
        <templateId root="2.16.840.1.113883.10.20.20.1.4"
assigningAuthorityName="GTR Cytogenetics Section"/>
        <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
codeSystemName="LOINC" displayName="Cytogenetics Section"/>
      </section>
    </structuredBody>
  </component>
</ClinicalDocument>
```
FINDINGS SECTION

The Findings Section is a narrative-only section. It resides within the TestDetailsSection and describes a specific finding of genetic testing. Note that the structured data is represented in a ClinicalGenomicStatement sub-templates nesting in the Test Details Section sub-templates.

1. SHALL conform to CDA Section
2. SHALL contain [1..1] code/@code = "TBD" Genetic Testing Findings Section (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-47)
3. SHALL contain [1..1] title = "Findings" (CONF-GTR-48)

Follow Up Genetic Tests Section

The FollowUpGeneticTestsSection section resides within the RecommendationSection (within SummarySection) and consists of tests to be performed in order to continue with the process. As much as possible, the section should contain structured entries representing the follow up tests, in a similar way of the performed tests representation in this report.

1. SHALL conform to CDA Section
2. **SHALL** contain [1..1] code/@code = "TBD" *Follow Up Genetic Tests Section* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-37)

3. **SHALL** contain [1..1] title = "Follow Up Genetic Tests" (CONF-GTR-38)

```xml
<?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">
  <component>
    <structuredBody>
      <component>
        <section>
          <templateId root="2.16.840.1.113883.10.20.20.1.1.3" assigningAuthorityName="GTR Follow Up Genetic Tests Section"/>
          <code code="TBD" codeSystem="2.16.840.1.113883.6.1" codeSystemName="LOINC" displayName="Follow Up Genetic Tests Section"/>
          <title>Follow Up Genetic Tests Section</title>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

*Figure 9: Follow Up Genetic Tests Section example*

### Follow Up Visits To Specialists Section

**[Section: templateId 2.16.840.1.113883.10.20.20.1.1.4]**

The FollowUpVisitsToSpecialists Section is a narrative-only section and resides within the RecommendationSection (within SummarySection).

1. **SHALL** conform to CDA Section


3. **SHALL** contain [1..1] title = "Follow Up Visits To Specialists" (CONF-GTR-40)

```xml
<?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">
  <component>
    <structuredBody>
      <component>
        <section>
          <templateId root="2.16.840.1.113883.10.20.20.1.1.4" assigningAuthorityName="GTR Follow Up Visits To Specialists Section"/>
          <code code="TBD" codeSystem="2.16.840.1.113883.6.1" codeSystemName="LOINC" displayName="Genetic Testing Follow Up Visits to Specialists"/>
          <title>Genetic Testing Follow Up Visits to Specialists</title>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

*Figure 10: Follow Up Visits To Specialists Section example*

### Gene Expression Section

**[Section: templateId 2.16.840.1.113883.10.20.20.1.3]**

The GeneExpressionSection resides at the highest level of the Genetic Testing Report and consists of data related to gene expression levels.
1. **SHALL** conform to *GTR Test Details Section* template (templateId: 2.16.840.1.113883.10.20.20.1.8)
2. **SHALL** contain [1..1] code/@code = "TBD" *Gene Expression Section* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-29)
3. **SHALL** contain [1..1] title = "Gene Expression" (CONF-GTR-30)
4. **SHOULD** contain [0..*] entry, such that it
   a. has @typeCode="COMP" COMP (component)
   b. contains *GTR Clinical Genomic Statement Gene Expression* (templateId: 2.16.840.1.113883.10.20.20.2.3)

<?xml version="1.0" encoding="UTF-8"?>
<ClinicalDocument xmlns:xsi="http://www.w3.org/2001/XMLSchema-instance"
xmllns="urn:hl7-org:v3" xsi:schemaLocation="urn:hl7-org:v3 CDA.xsd">
    <component>
        <structuredBody>
            <component>
                <section>
                    <templateId root="2.16.840.1.113883.10.20.20.1.8"
                        assigningAuthorityName="GTR Test Details Section"/>
                    <templateId root="2.16.840.1.113883.10.20.20.1.3"
                        assigningAuthorityName="GTR Gene Expression Section"/>
                    <code codeSystem="LOINC" codeSystemName="LOINC" displayName="Gene Expression Section"/>
                    <title>Gene Expression Section</title>
                    <entry>
                        <observation classCode="OBS" moodCode="EVN">
                            <templateId root="2.16.840.1.113883.10.20.20.2.1"
                                assigningAuthorityName="GTR Clinical Genomic Statement"/>
                            <templateId root="2.16.840.1.113883.10.20.20.2.3"
                                assigningAuthorityName="GTR Clinical Genomic Statement Gene Expression"/>
                            <code codeSystemName="HUGO Gene Names"/>
                            <methodCode/>
                        </observation>
                    </entry>
                    </section>
                </component>
            </structuredBody>
        </component>
    </ClinicalDocument>

Figure 11: Gene Expression Section example

**Genetic Variations Section**

[Section: templateId 2.16.840.1.113883.10.20.20.1.2]

The GeneticVariationSection resides at the highest level of the Genetic Testing Report and consists of data related to genetic variations. The typical genetic variation described in this section is in the order of variations occurring in a gene. It should not cover cytogenetic changes for example.

1. **SHALL** conform to *GTR Test Details Section* template (templateId: 2.16.840.1.113883.10.20.20.1.8)
2. **SHALL** contain [1..1] code/@code = "TBD" *Genetic Variations Section* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-31)
3. **SHALL** contain [1..1] title = "Genetic Variations" (CONF-GTR-32)
4. **SHOULD** contain [0..*] entry, such that it
   a. has @typeCode="COMP" COMP (component)
   b. contains *GTR Clinical Genomic Statement Genetic Variation* (templateId: 2.16.840.1.113883.10.20.20.2.1)

<?xml version="1.0" encoding="UTF-8"?>
<ClinicalDocument xmlns:xsi="http://www.w3.org/2001/XMLSchema-instance"
xmllns="urn:hl7-org:v3" xsi:schemaLocation="urn:hl7-org:v3 CDA.xsd">
Indications Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.11]

The IndicationSection nests within the TestDetailsSection and its text attribute consists of narrative describing the indication of performing the genetic tests. It should also consist of structured indication observations that can be referenced from Clinical Genomic Statement template.

1. SHALL conform to CDA Section
2. SHALL contain [1..1] code/@code = "TBD" Genetic Testing Indications Section (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-10)
3. SHALL contain [1..1] title = "Indications" (CONF-GTR-11)
4. Contains [0..*] entry, such that it
   a. contains GTR Indication Observation (templateId: 2.16.840.1.113883.10.20.20.3.3)
Interpretation Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.13]

The InterpretationSection is a narrative-only section. It nests within the TestDetailsSection and its text attribute consists of narrative describing the interpretation of the genetic test at stake. Note that structured representation of the interpretation is part of the Clinical Genomic Statement template.

1. SHALL conform to CDA Section
2. SHALL contain [1..1] code/@code = "TBD" Genetic Testing Interpretation Section (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-20)
3. SHALL contain [1..1] title = "Interpretation" (CONF-GTR-21)

```xml
<?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">
  <component>
    <structuredBody>
      <component>
        <section>
          <templateId root="2.16.840.1.113883.10.20.20.1.13"
                      assigningAuthorityName="GTR Interpretation Section"/>
          <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
                codeSystemName="LOINC" displayName="Genetic Testing Interpretation Section"/>
          <title>Genetic Testing Interpretation Section</title>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

Figure 14: Interpretation Section example

Methodology Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.9.2]

The MethodologySection is a narrative-only section. It nests within the TestInformationSection and its text attribute consists of narrative describing methodology of the genetic test at stake.

1. SHALL conform to CDA Section
2. SHALL contain [1..1] code/@code = "TBD" Genetic Testing Methodology (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-16)
3. SHALL contain [1..1] title = "Methodology" (CONF-GTR-17)

```xml
<?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">
  <component>
    <structuredBody>
      <component>
        <section>
          <templateId root="2.16.840.1.113883.10.20.20.1.9.2"
                      assigningAuthorityName="GTR Methodology Section"/>
          <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
                codeSystemName="LOINC" displayName="Genetic Testing Methodology"/>
          <title>Genetic Testing Methodology</title>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```
Other Testing Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.6]

A 'catcher' section of genomic observations that could not be represented in one of the specialized sections such as genetic variation or gene expression. It is encouraged to use codified tests, i.e., represent the test details in structured entries with recognized vocabularies.

1. SHALL conform to GTR Test Details Section template (templateId: 2.16.840.1.113883.10.20.20.1.8)
3. SHALL contain [1..1] title = "Other Testing" (CONF-GTR-28)
4. SHOULD contain [0..*] entry, such that it
   a. has @typeCode="COMP" COMP (component)
   b. contains GTR Clinical Genomic Statement (templateId: 2.16.840.1.113883.10.20.20.2)

Overall Interpretation Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.1.1]

The Overall Interpretive Section describes the overall interpretation of the genetic test performed. It is further specialized by its sub-templates that represent overall interpretation by various testing types. Note that its own code and value could potentially represent the overall interpretation of multiple overall interpretations in case the report describes multiple tests performed (e.g., genetic variation and gene expression tests).

1. SHALL conform to CDA Section
2. **SHALL** contain [1..1] `code/@code = "TBD"` *Genetic Testing Overall Interpretation Section* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-35)

3. **SHALL** contain [1..1] `title = "Overall Interpretation"` (CONF-GTR-36)

4. **MAY** contain [0..1] `entry`, such that it
   a. has `@typeCode="COMP"` COMP (component)
   b. contains *GTR Overall Interpretive Phenotype Observation Pharmacogenomic Drug Efficacy* (templateId: 2.16.840.1.113883.10.20.20.2.5.7)

5. **MAY** contain [0..1] `entry`, such that it
   a. has `@typeCode="COMP"` COMP (component)
   b. contains *GTR Overall Interpretive Phenotype Observation Pharmacogenomic Drug Metabolism* (templateId: 2.16.840.1.113883.10.20.20.2.5.8)

6. **MAY** contain [0..1] `entry`, such that it
   a. has `@typeCode="COMP"` COMP (component)
   b. contains *GTR Overall Interpretive Phenotype Observation Genetic Disease* (templateId: 2.16.840.1.113883.10.20.20.2.5.5)

7. **MAY** contain [0..1] `entry`, such that it
   a. has `@typeCode="COMP"` COMP (component)
   b. contains *GTR Overall Interpretive Phenotype Observation Genetic Disease Carrier* (templateId: 2.16.840.1.113883.10.20.20.2.5.6)

8. Contains [0..1] `entry`, such that it
   a. contains *GTR Overall Interpretive Phenotype Observation Chromosome Analysis* (templateId: 2.16.840.1.113883.10.20.20.2.5.4.2)
Figure 17: Overall Interpretation Section example

**Recommendations Section**

The RecommendationsSection is a narrative-only section. It nests within the SummarySection and its text attribute consists of narrative describing recommended actions such as follow-up genetic testing etc..

1. SHALL conform to CDA Section
3. SHALL contain [1..1] title = "Recommendations" (CONF-GTR-86)
4. MAY contain [0..1] component, such that it
   a. contains GTR Follow Up Genetic Tests Section (templateId: 2.16.840.1.113883.10.20.20.1.1.3)
5. MAY contain [0..1] component, such that it
   a. contains GTR Follow Up Visits To Specialists Section (templateId: 2.16.840.1.113883.10.20.20.1.1.4)
6. MAY contain [0..1] component, such that it
   a. contains GTR Recommended Actions Section (templateId: 2.16.840.1.113883.10.20.20.1.1.5)
Recommended Actions Section

A 'catcher' section of recommended actions that could not be represented in one of the specialized sections nesting under "Recommendations Section". It is encouraged to use codified recommended actions as much as possible, e.g., use procedure codes if recommended actions are type of procedures.

1. SHALL conform to CDA Section
2. Contains [1..1] code
3. SHALL contain [1..1] title = "Recommended Actions"

References Section

The ReferencesSection section consists of references to scientific literature that supports the description of the test. It nests within the TestInformationSection and its text attribute consists of narrative describing scientific references of the genetic test at stake, and optionally structured entries representing publications identified through common ids like PubMed ids.

1. SHALL conform to CDA Section
2. SHALL contain [1..1] code/@code = "TBD" Genetic Testing References (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-14)
3. **SHALL** contain [1..1] title = "References" (CONF-GTR-15)

```xml
<?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">
  <component>
    <structuredBody>
      <component>
        <section>
          <templateId root="2.16.840.1.113883.10.20.20.1.9.3"
assigningAuthorityName="GTR References Section"/>
          <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
codeSystemName="LOINC" displayName="Genetic Testing References"/>
          <title>Genetic Testing References</title>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

Figure 20: References Section example

**Specimen Section**

[Section: templateId 2.16.840.1.113883.10.20.20.1.7]

The Specimen Section describes the specimen used for the genetic testing at stake and the genomic source class. This is narrative-only section because the structured data describing the specimen and source calls are part of the ClinicalGenomicStatement template.

1. **SHALL** conform to CDA Section
2. **SHALL** contain [1..1] code/@code = "TBD" (Code system: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-33)
3. **SHALL** contain [1..1] title = "Specimen and Genomic Source Class" (CONF-GTR-34)

```xml
<?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">
  <component>
    <structuredBody>
      <component>
        <section>
          <templateId root="2.16.840.1.113883.10.20.20.1.7"
assigningAuthorityName="GTR Specimen Section"/>
          <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
codeSystemName="LOINC" displayName="Genetic Testing Specimen and Genomic Source Section"/>
          <title>Genetic Testing Specimen and Genomic Source Section</title>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

Figure 21: Specimen Section example

**Summary Section**

[Section: templateId 2.16.840.1.113883.10.20.20.1.1]

The Summary Section resides at the highest level of the Genetic Testing Report and consists of several sub-sections describing the overall interpretation of the various genetic tests described in the GTR as well as the genomic source type, recommended follow-up genetic tests, specialist visits, and care plan.
1. SHALL conform to CDA Section
2. SHALL contain [1..1] code/@code = "TBD" Genetic Testing Summary Section (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-43)
3. SHALL contain [1..1] title = "Summary" (CONF-GTR-44)
4. Contains [0..*] component, such that it
   a. contains GTR Overall Interpretation Section (templateId: 2.16.840.1.113883.10.20.1.1.1)
5. Contains [0..1] component, such that it
   a. contains GTR Tests Performed Section (templateId: 2.16.840.1.113883.10.20.1.10)
6. Contains [0..1] component, such that it
   a. contains GTR Test Information Section (templateId: 2.16.840.1.113883.10.20.1.9)
7. Contains [0..1] component, such that it
   a. contains GTR Recommendations Section (templateId: 2.16.840.1.113883.10.20.1.1.5)

```xml
<?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">
  <component>
    <structuredBody>
      <component>
        <section>
          <templateId root="2.16.840.1.113883.10.20.1.1" assigningAuthorityName="GTR Summary Section"/>
          <code code="TBD" codeSystem="2.16.840.1.113883.6.1" codeSystemName="LOINC" displayName="Genetic Testing Summary Section"/>
          <title>Summary</title>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

Figure 22: Summary Section example

### Test Details Section

[Section: templateId 2.16.840.1.113883.10.20.1.8]

The Test Details Section is the blueprint for all specialized sections that appear on the GTR top level such as the genetic variation section, cytogenetic section, etc. It consists of those sub-sections considered to appear in the same way in each of the specialized sections, e.g., specimen, indications, tests performed, and test information. Note that the interpretation section is narrative only and the structured interpretation appears as part of the clinical genomic statement.

1. SHALL conform to CDA Section
2. Contains [0..1] code
3. Contains [0..1] title
4. SHOULD contain [0..1] component, such that it
   a. contains GTR Specimen Section (templateId: 2.16.840.1.113883.10.20.1.7)
5. SHOULD contain [0..1] component, such that it
   a. contains GTR Indications Section (templateId: 2.16.840.1.113883.10.20.1.11)
6. SHOULD contain [0..1] component, such that it
   a. contains GTR Tests Performed Section (templateId: 2.16.840.1.113883.10.20.1.10)
7. SHOULD contain [0..1] component, such that it
   a. contains GTR Findings Section (templateId: 2.16.840.1.113883.10.20.1.12)
8. SHOULD contain [0..1] component, such that it 
   a. contains GTR Interpretation Section (templateId: 2.16.840.1.113883.10.20.1.13)
9. SHOULD contain [0..1] component, such that it 
   a. contains GTR Test Information Section (templateId: 2.16.840.1.113883.10.20.1.9)
Test Information Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.9]

The TestInformationSection is a narrative-only section. It nests within the TestDetailsSection and its sub-sections consist of narrative describing information on the genetic test at stake.

1. SHALL conform to CDA Section
3. SHALL contain [1..1] title (CONF-GTR-46)
4. SHOULD contain [0..1] component, such that it
   a. contains GTR Background Section (templateId: 2.16.840.1.113883.10.20.20.1.9.1)
5. SHOULD contain [0..1] component, such that it
   a. contains GTR Methodology Section (templateId: 2.16.840.1.113883.10.20.20.1.9.2)
6. SHOULD contain [0..1] component, such that it
   a. contains GTR References Section (templateId: 2.16.840.1.113883.10.20.20.1.9.3)
Tests Performed Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.10]

The TestsPerformedSection nests within the TestDetailsSection and its text attribute consists of narrative describing the tests performed including those which did not have any results. It can consist of observations describing the performed tests.

1. SHALL conform to CDA Section
2. SHALL contain [1..1] code/@code = "TBD" Genetic Tests Performed Section (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-12)
3. SHALL contain [1..1] title = "Tests Performed" (CONF-GTR-13)
4. SHOULD contain [0..*] entry, such that it
   a. has @typeCode="COMP" COMP (component)
   b. contains GTR Test Performed Observation (templateId: 2.16.840.1.113883.10.20.20.3.4)
Figure 25: Tests Performed Section example
This section of the Implementation Guide details the clinical statement entries referenced in the document section templates. The clinical statement entry templates are arranged alphabetically.
• Interpretive Phenotype Observation Genetic Variation
• Interpretive Phenotype Observation Pharmacogenomic
• Interpretive Phenotype Observation Pharmacogenomic Drug Efficacy
• Interpretive Phenotype Observation Pharmacogenomic Drug Metabolism
• Overall Interpretive Phenotype Observation Chromosome Analysis
• Overall Interpretive Phenotype Observation Genetic Disease
• Overall Interpretive Phenotype Observation Genetic Disease Carrier
• Overall Interpretive Phenotype Observation Pharmacogenomic Drug Efficacy
• Overall Interpretive Phenotype Observation Pharmacogenomic Drug Metabolism
• Test Performed Observation
Clinical Genomic Statement

[Observation: templateId 2.16.840.1.113883.10.20.20.2]

The ClinicalGenomicStatement template is the core of the structured portion of the GTR Implementation Guide. It is constituted of a genetic observation, e.g., a genetic variation, which can be associated with an indication to performing this genetic observation, as well as an association with the interpretation of that observation. Due to the complexity of the interpretation of genetic observations, this template disallows the use of the interpretationCode attribute, and rather uses an association to InterpretivePhenotypeObservation. Nevertheless, due to the specificity of the interpretation, sub-templates of this template further constrain it by using InterpretivePhenotypeObservation sub-templates. For example, ClinicalGenomicStatementGeneticVariation is a sub-template of ClinicalGenomicStatement and is associated with InterpretivePhenotypeObservationGeneticVariation which is a sub-template of InterpretivePhenotypeObservation.

1. SHALL conform to CDA Observation
2. SHALL contain [1..1] code (CONF-GTR-9)
3. Contains [0..1] value
4. SHALL contain [0..1] entryRelationship, such that it
   a. has @typeCode="RSON" RSON (has reason)
   b. contains GTR Indication Observation (templateId: 2.16.840.1.113883.10.20.20.3.3)
5. SHALL contain [0..1] entryRelationship, such that it
   a. has @typeCode="MFST" MFST (is manifestation of)
   b. contains GTR Interpretive Phenotype Observation (templateId: 2.16.840.1.113883.10.20.20.2.5)
6. SHOULD contain [0..*] methodCode
7. SHALL contain [0..1] entryRelationship, such that it
   a. has @typeCode="COMP" COMP (has component)
   b. contains GTR Genomic Source Class (templateId: 2.16.840.1.113883.10.20.20.3.2)
8. Contains [0..1] performer, such that it
   a. contains GTR Performer (templateId: 2.16.840.1.113883.10.20.20.3.2)
9. Contains [0..1] geneticSpecimen, such that it
   a. contains GTR Genetic Specimen (templateId: 2.16.840.1.113883.10.20.20.3.1)
10. SHALL satisfy: GTR Observations SHALL NOT use the interpretationCode attribute, rather use the InterpretivePhenotypeObservation template and its sub-templates. (CONF-GTR-8)
Clinical Genomic Statement Cytogenetics

[Observation: templateId 2.16.840.1.113883.10.20.20.2.2]

The ClinicalGenomicStatementCytogenetics template is a sub-template of ClinicalGenomicStatement. It is used by the CytogeneticSection to carry the structured data in that section. It further constrains the InterpretivePhenotypeObservation abstract template by associating to the InterpretivePhenotypeObservationCytogenetics.

1. SHALL conform to *GTR Clinical Genomic Statement* template (templateId: 2.16.840.1.113883.10.20.20.2)

2. SHALL contain [1..1] code/@code = "62356-1" Chromosome analysis result in ISCN expression in Blood or Tissue by Molecular genetics method (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-69)

3. SHALL contain [0..1] value (CONF-GTR-70)

4. Contains [0..1] entryRelationship, such that it
   a. contains *GTR Interpretive Phenotype Observation Cytogenetics* (templateId: 2.16.840.1.113883.10.20.20.2.5.1)

5. Contains [0..1] entryRelationship, such that it
   a. contains *GTR Clinical Genomic Statement Cytogenetics Cells Count* (templateId: 2.16.840.1.113883.10.20.20.2.2.2)

6. Contains [0..1] entryRelationship, such that it
   a. contains *GTR Clinical Genomic Statement Cytogenetics Cells Analyzed Count* (templateId: 2.16.840.1.113883.10.20.20.2.2.1)

7. Contains [0..1] entryRelationship, such that it
   a. contains *GTR Clinical Genomic Statement Cytogenetics Colonies Count* (templateId: 2.16.840.1.113883.10.20.20.2.2.4)

8. Contains [0..1] entryRelationship, such that it
   a. contains *GTR Clinical Genomic Statement Cytogenetics Cells Karyotyped Count* (templateId: 2.16.840.1.113883.10.20.20.2.2.3)
9. Contains [0..1] entryRelationship, such that it
   a. contains GTR Clinical Genomic Statement Cytogenetics ISCN Band Level (templateId:
      2.16.840.1.113883.10.20.2.2.5)
10. SHALL satisfy: value SHALL be assigned a string composed using the expression syntax of International System
    for Human Cytogenetics Nomenclature (ISCN).

```xml
<?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">
  <component>
    <structuredBody>
      <component>
        <section>
          <entry>
            <observation classCode="OBS" moodCode="EVN">
              <templateId root="2.16.840.1.113883.10.20.20.2.1"
                assigningAuthorityName="GTR Clinical Genomic Statement"/>
              <templateId root="2.16.840.1.113883.10.20.20.2.2"
                assigningAuthorityName="GTR Clinical Genomic Statement Cytogenetics"/>
              <code/>
              <methodCode/>
              <entryRelationship>
                <observation classCode="OBS" moodCode="EVN">
                  <templateId root="2.16.840.1.113883.10.20.20.3.3"
                    assigningAuthorityName="GTR Indication Observation"/>
                  <code code="MTHU008863" codeSystem="2.16.840.1.113883.6.1" codeSystemName="LOINC" displayName="Indications description"/>
                  <value xsi:type="CD"/>
                </observation>
              </entryRelationship>
              <entryRelationship>
                <observation classCode="OBS" moodCode="EVN">
                  <templateId root="2.16.840.1.113883.10.20.20.2.5"
                    assigningAuthorityName="GTR Interpretive Phenotype Observation"/>
                  <value xsi:type="CD"/>
                  <methodCode/>
                </observation>
              </entryRelationship>
              <entryRelationship>
                <observation classCode="OBS" moodCode="EVN">
                  <templateId root="2.16.840.1.113883.10.20.20.2.1"
                    assigningAuthorityName="GTR Clinical Genomic Statement"/>
                  <templateId root="2.16.840.1.113883.10.20.20.3.2"
                    assigningAuthorityName="GTR Genomic Source Class"/>
                  <code/>
                  <value xsi:type="CD"/>
                  <methodCode/>
                </observation>
              </entryRelationship>
            </observation>
          </entry>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

Figure 27: Clinical Genomic Statement Cytogenetics example
Clinical Genomic Statement Cytogenetics Cells Analyzed Count

[Observation: templateId 2.16.840.1.113883.10.20.20.2.2.1]

The ClinicalGenomicStatementCytogeneticsCellsAnalyzedCount template is a sub-template of ClinicalGenomicStatementCytogenetics and is used to carry the no. of cells analyzed in a cytogenetics test.

1. SHALL conform to GTR Clinical Genomic Statement template (templateId: 2.16.840.1.113883.10.20.20.2)
2. MAY contain [1..1] code/@code = "62360-3" Cells analyzed [#] in Blood or Tissue by Molecular genetics method (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26)
3. SHALL contain [0..1] value, where its data type is INT

Figure 28: Clinical Genomic Statement Cytogenetics Cells Analyzed Count example

Clinical Genomic Statement Cytogenetics Cells Count

[Observation: templateId 2.16.840.1.113883.10.20.20.2.2.2]

The ClinicalGenomicStatementCytogeneticsCellsCount template is a sub-template of ClinicalGenomicStatementCytogenetics and is used to carry the no. of cells counted in a cytogenetics test.

1. SHALL conform to GTR Clinical Genomic Statement template (templateId: 2.16.840.1.113883.10.20.20.2)
2. MAY contain [1..1] code/@code = "62361-1" Cells counted [#] in Blood or Tissue by Molecular genetics method (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26)
3. SHALL contain [0..1] value, where its data type is INT

Figure 29: Clinical Genomic Statement Cytogenetics Cells Count example

Clinical Genomic Statement Cytogenetics Cells Karyotyped Count

[Observation: templateId 2.16.840.1.113883.10.20.20.2.2.3]

The ClinicalGenomicStatementCytogeneticsCellsKaryotypedCount template is a sub-template of ClinicalGenomicStatement and is used to carry the no. of cells karyotyped in a cytogenetics test.

1. SHALL conform to GTR Clinical Genomic Statement template (templateId: 2.16.840.1.113883.10.20.20.2)
2. MAY contain [1..1] code/@code = "55199-4" Cells karyotyped.total [#] in Blood (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26)
3. SHALL contain [0..1] value, where its data type is INT

Figure 30: Clinical Genomic Statement Cytogenetics Cells Karyotyped Count example

Clinical Genomic Statement Cytogenetics Colonies Count

[Observation: templateId 2.16.840.1.113883.10.20.20.2.2.4]

The ClinicalGenomicStatementCytogeneticsColoniesCount template is a sub-template of ClinicalGenomicStatement and is used to carry the no. of colonies counted a cytogenetics test. Colony is a discrete focus of cells that is harvested and stained while attached to the cell culture growth substrate.

1. SHALL conform to GTR Clinical Genomic Statement template (templateId: 2.16.840.1.113883.10.20.20.2)
2. MAY contain [1..1] code/@code = "62362-9" Colonies counted [#] in Blood or Tissue by Molecular genetics method (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26)
3. **SHALL** contain [0..1] value, where its data type is INT

**Figure 31: Clinical Genomic Statement Cytogenetics Colonies Count example**

**Clinical Genomic Statement Cytogenetics ISCN Band Level**

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5]

The ClinicalGenomicStatementCytogeneticsISCNBandLevel template is a sub-template of ClinicalGenomicStatement and is used to carry the ISCN band level of the cytogenetics test.

1. **SHALL** conform to *GTR Clinical Genomic Statement* template (templateId: 2.16.840.1.113883.10.20.20.2)
2. **MAY** contain [1..1] code/@code = "62358-7" ISCN band level (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26)
3. **SHALL** contain [0..1] value, which **SHALL** be selected from ValueSet 2.16.840.1.113883.10.20.20.9.13 ISCN band level STATIC, where its data type is CD
4. **SHALL** satisfy: GTR ClinicalGenomicStatementCytogeneticsISCNBandLevel (self) SHALL satisfy: If self.code.code=62358-7 (LOINC code for ISCN band level), then self.value@code SHALL be drawn from the LOINC Value Set 2.16.840.1.113883.10.20.20.9.13 (ISCN band level in Blood or Tissue by Molecular genetics: 62358-7).

**Figure 32: Clinical Genomic Statement Cytogenetics ISCN Band Level example**

**Clinical Genomic Statement Gene Expression**

[Observation: templateId 2.16.840.1.113883.10.20.20.2.3]

The ClinicalGenomicStatementGeneExpression template is a sub-template of ClinicalGenomicStatement. It is used by the GeneExpressionSection to carry the structured data in that section. It further constrains the InterpretivePhenotypeObservation abstract template by associating to the InterpretivePhenotypeObservationGeneExpression. This template is not constrained as the genetic variation and cytogenetics are, due to the rapid developments in the field of gene expression. As much as possible, assign commonly-used codes in the gene expression field into the code and value attributes of this template.

1. **SHALL** conform to *GTR Clinical Genomic Statement* template (templateId: 2.16.840.1.113883.10.20.20.2)
2. **SHOULD** contain [1..1] code (CodeSystem: HUGO Gene Names STATIC) (CONF-GTR-68)
3. Contains [0..1] value
4. Contains [0..1] entryRelationship, such that it
   a. contains *GTR Interpretive Phenotype Observation Gene Expression* (templateId: 2.16.840.1.113883.10.20.20.2.5.2)
5. Contains [0..1] entryRelationship, such that it
   a. contains *GTR Interpretive Phenotype Observation Pharmacogenomic Drug Efficacy* (templateId: 2.16.840.1.113883.10.20.20.2.5.4.1)
6. Contains [0..1] entryRelationship, such that it
   a. contains *GTR Interpretive Phenotype Observation Pharmacogenomic Drug Metabolism* (templateId: 2.16.840.1.113883.10.20.20.2.5.4.2)
Clinical Genomic Statement Genetic Variation

[Observation: templateId 2.16.840.1.113883.10.20.20.2.1]

The ClinicalGenomicStatementGeneticVariation template is a sub-template of ClinicalGenomicStatement. It is used by the GeneticVariationSection to carry the structured data in that section. It is associated with InterpretivePhenotypeObservation sub-templates.

1. SHALL conform to GTR Clinical Genomic Statement template (templateId: 2.16.840.1.113883.10.20.20.2)

2. SHALL contain [1..1] code (CONF-GTR-90)

   - In principle, the code attribute should designate the type of the genetic variation being described in this Clinical Genomic Statement. Typically, a genetic variation can be characterized by multiple aspects, e.g., DNA change, amino acid change, Transcript Reference Sequence Identifier, etc. It is important to note that there is no standard for any kind of genetic variation and even HGVS nomenclature doesn't cover all cases. Also, 'gene-centric' variation notations might be disadvantageous because in some genomic locations, a variant may be influencing several genes (or transcripts of the same gene) and may have different effects, for example, an indel in an intron of one transcript may be a frame shift in an exon another transcript for the same gene.
When possible, a coded panel of such characteristics should be used, for example, the LOINC panel "DNA Analysis Discrete Sequence Variant Panel" (code=55208-3) designed for clinical environment. When this code is assigned to the code attribute, then this Clinical Genomic Statement SHALL consist of nesting observations describing the Gene Identifier, Transcript Reference Sequence Identifier, DNA Sequence Variation, DNA Sequence Variation Type, Amino Acid Change, Amino Acid Change Type, DNA Region Name, Allelic State, Genomic Source Class (if not specified already in the Summary Section). The constraining of these nesting observations are described in detail in the association of this Clinical Genomic Statement, including their code and binding value sets. If code is not assigned with the above-mentioned LOINC Panel, then it should use either the Human Genome Variation Society (HGVS) nomenclature or other recognized notation of genetic variations (TBD).

3. **SHOULD** contain [0..1] value (CONF-GTR-55)
4. **MAY** contain [0..1] entryRelationship, such that it  
   a. has @typeCode="SUBJ" SUBJ (has subject)  
   b. contains *GTR Clinical Genomic Statement Genetic Variation Amino Acid Change* (templateId: 2.16.840.1.113883.10.20.2.1.1) (CONF-GTR-92)
5. **MAY** contain [0..1] entryRelationship, such that it  
   a. has @typeCode="SUBJ" SUBJ (has subject)  
   b. contains *GTR Clinical Genomic Statement Genetic Variation DNA Change* (templateId: 2.16.840.1.113883.10.20.2.1.2) (CONF-GTR-93)
6. **MAY** contain [0..1] entryRelationship, such that it  
   a. has @typeCode="SUBJ" SUBJ (has subject)  
   b. contains *GTR Clinical Genomic Statement Genetic Variation Zygosity* (templateId: 2.16.840.1.113883.10.20.2.1.4) (CONF-GTR-94)
7. **MAY** contain [0..1] entryRelationship, such that it  
   a. has @typeCode="SUBJ" SUBJ (has subject)  
   b. contains *GTR Clinical Genomic Statement Genetic Variation DNA Region Name* (templateId: 2.16.840.1.113883.10.20.2.1.3) (CONF-GTR-95)
8. **MAY** contain [0..1] entryRelationship, such that it  
   a. has @typeCode="SPRT" SPRT (has support)  
   b. contains *GTR Interpretive Phenotype Observation Genetic Variation* (templateId: 2.16.840.1.113883.10.20.2.5.3) (CONF-GTR-96)
9. **MAY** contain [0..1] entryRelationship, such that it  
   a. has @typeCode="SPRT" SPRT (has support)  
   b. contains *GTR Interpretive Phenotype Observation Pharmacogenomic Drug Efficacy* (templateId: 2.16.840.1.113883.10.20.2.5.4.1) (CONF-GTR-97)
10. **MAY** contain [0..1] entryRelationship, such that it  
    a. has @typeCode="SPRT" SPRT (has support)  
    b. contains *GTR Interpretive Phenotype Observation Pharmacogenomic Drug Metabolism* (templateId: 2.16.840.1.113883.10.20.2.5.4.2) (CONF-GTR-98)
11. **SHALL** satisfy: If code=55208-3 (LOINC code for "DNA Analysis Discrete Sequence Variant Panel"), then value SHALL NOT be used. (CONF-GTR-91)
<templateId root="2.16.840.1.113883.10.20.20.2.3"
  assigningAuthorityName="GTR Clinical Genomic Statement Gene Expression"/>
<code/>
</entryRelationship>
<entryRelationship>
  <observation classCode="OBS" moodCode="EVN">
    <templateId root="2.16.840.1.113883.10.20.20.2.1"
      assigningAuthorityName="GTR Clinical Genomic Statement"/>
    <templateId root="2.16.840.1.113883.10.20.20.2.3.1"
      assigningAuthorityName="GTR Clinical Genomic Statement Genetic Variation Amino Acid Change"/>
    <code/>
    <value xsi:type="CD"/>
    <methodCode/>
  </observation>
</entryRelationship>
<entryRelationship>
  <observation classCode="OBS" moodCode="EVN">
    <templateId root="2.16.840.1.113883.10.20.20.2.1"
      assigningAuthorityName="GTR Clinical Genomic Statement"/>
    <templateId root="2.16.840.1.113883.10.20.20.2.3.2"
      assigningAuthorityName="GTR Clinical Genomic Statement Genetic Variation DNA Change"/>
    <code/>
    <value xsi:type="CD"/>
    <methodCode/>
  </observation>
</entryRelationship>
<entryRelationship>
  <observation classCode="OBS" moodCode="EVN">
    <templateId root="2.16.840.1.113883.10.20.20.2.1"
      assigningAuthorityName="GTR Clinical Genomic Statement"/>
    <templateId root="2.16.840.1.113883.10.20.20.2.3.3"
      assigningAuthorityName="GTR Clinical Genomic Statement Genetic Variation Zygosity"/>
    <code/>
    <value xsi:type="CD"/>
    <methodCode/>
  </observation>
</entryRelationship>
<entryRelationship>
  <observation classCode="OBS" moodCode="EVN">
    <templateId root="2.16.840.1.113883.10.20.20.2.1"
      assigningAuthorityName="GTR Clinical Genomic Statement"/>
    <templateId root="2.16.840.1.113883.10.20.20.2.3.4"
      assigningAuthorityName="GTR Clinical Genomic Statement Genetic Variation DNA Region Name"/>
    <code/>
    <value xsi:type="ST"/>
    <methodCode/>
  </observation>
</entryRelationship>
<entryRelationship>
  <observation classCode="OBS" moodCode="EVN">
    <templateId root="2.16.840.1.113883.10.20.20.2.5"
      assigningAuthorityName="GTR Interpretive Phenotype Observation"/>
    <templateId root="2.16.840.1.113883.10.20.20.2.5.3"
      assigningAuthorityName="GTR Interpretive Phenotype Observation Genetic Variation"/>
    <code/>
    <value xsi:type="CD"/>
    <methodCode/>
  </observation>
</entryRelationship>
<observation classCode="OBS" moodCode="EVN">
    <templateId root="2.16.840.1.113883.10.20.2.5">
        assigningAuthorityName="GTR Interpretive Phenotype Observation"/>
    <templateId root="2.16.840.1.113883.10.20.2.5.4">
        assigningAuthorityName="GTR Interpretive Phenotype Observation Pharmacogenomic"/>
    </observation>
</observation>

<observation classCode="OBS" moodCode="EVN">
    <templateId root="2.16.840.1.113883.10.20.2.5">
        assigningAuthorityName="GTR Interpretive Phenotype Observation Pharmacogenomic"/>
    <templateId root="2.16.840.1.113883.10.20.2.5.4.1">
        code code="51961-1" codeSystem="2.16.840.1.113883.6.1" codeSystemName="LOINC" displayName="Drug efficacy sequence variation interpretation"/>
    </observation>
</observation>

<observation classCode="OBS" moodCode="EVN">
    <templateId root="2.16.840.1.113883.10.20.2.5">
        assigningAuthorityName="GTR Interpretive Phenotype Observation Pharmacogenomic"/>
    <templateId root="2.16.840.1.113883.10.20.2.5.4.2">
        code code="53040-2" codeSystem="2.16.840.1.113883.6.1" codeSystemName="LOINC" displayName="Drug metabolism sequence variation interpretation"/>
    </observation>
</observation>

<observation classCode="OBS" moodCode="EVN">
    <templateId root="2.16.840.1.113883.10.20.2.5.3">
        assigningAuthorityName="GTR Indication Observation"/>
    <code code="MTHU008863" codeSystem="2.16.840.1.113883.6.1" codeSystemName="LOINC" displayName="Indications description"/>
    </observation>
</observation>

<observation classCode="OBS" moodCode="EVN">
    <templateId root="2.16.840.1.113883.10.20.2.5">
        assigningAuthorityName="GTR Interpretive Phenotype Observation Pharmacogenomic"/>
    </observation>
</observation>

<observation classCode="OBS" moodCode="EVN">
    <templateId root="2.16.840.1.113883.10.20.2.5.2">
        assigningAuthorityName="GTR Clinical Genomic Statement"/>
    <templateId root="2.16.840.1.113883.10.20.2.1">
        assigningAuthorityName="GTR Genomic Source Class"/>
    </templateId root="2.16.840.1.113883.10.20.3.2">
        code/>
    <value xsi:type="CD"/>
    </observation>
</observation>
</observation>
</entryRelationship>
</entry Relationship>

</observation>
</structuredBody>
</component>
Clinical Genomic Statement Genetic Variation Amino Acid Change

[Observation: templateId 2.16.840.1.113883.10.20.20.2.1.1]

The ClinicalGenomicStatementGeneticVariationAminoAcid template is a sub-template of ClinicalGenomicStatement and is used to carry the amino acid change of that genetic variation.

1. SHALL conform to GTR Clinical Genomic Statement template (templateId: 2.16.840.1.113883.10.20.20.2)

2. MAY contain [1..1] code/@code = "48006-1" Amino acid change type (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-60)

3. MAY contain [0..1] value, which MAY be selected from ValueSet 2.16.840.1.113883.10.20.20.9.2 Amino acid change STATIC, where its data type is CD (CONF-GTR-61)

4. SHALL satisfy: GTR ClinicalGenomicStatementGeneticVariationAmino AcidChange (self) SHALL satisfy: If self.code@code=48006-1 (LOINC code for Amino acid change type), then self.value@code SHALL be drawn from the LOINC Value Set 2.16.840.1.113883.10.20.20.9.2 (Amino acid change type: 48006-1) (CONF-GTR-59)
Clinical Genomic Statement Genetic Variation DNA Change

[Observation: templateId 2.16.840.1.113883.10.20.20.2.1.2]

The ClinicalGenomicStatementGeneticVariationDNAChange template is a sub-template of ClinicalGenomicStatement and is used to carry the DNA change of that genetic variation.

1. SHALL conform to GTR Clinical Genomic Statement template (templateId: 2.16.840.1.113883.10.20.20.2)
2. MAY contain [1..1] code/@code = "48019-4" DNA sequence variation type (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-57)
3. MAY contain [0..1] value, which MAY be selected from ValueSet 2.16.840.1.113883.10.20.20.9.4 DNA sequence change STATIC, where its data type is CD (CONF-GTR-58)
4. SHALL satisfy: GTR ClinicalGenomicStatementGeneticVariationDNAChange (self) SHALL satisfy: If self.code/@code=48019-4 (LOINC code for DNA sequence variation type), then self.value.code SHALL be drawn from the LOINC Value Set 2.16.840.1.113883.10.20.20.9.4 (DNA sequence variation type: 48019-4). (CONF-GTR-56)
The ClinicalGenomicStatementGeneticVariationDNAChange template is a sub-template of ClinicalGenomicStatement and is used to carry the DNA change of that genetic variation.

1. **SHALL** conform to *GTR Clinical Genomic Statement* template (templateId: 2.16.840.1.113883.10.20.20.2)

2. **MAY** contain [1..1] code/@code = "47999-8" *DNA Region Name* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-102)

3. **MAY** contain [0..1] value, where its data type is ST

```xml
<ClinicalDocument xmlns:xsi="http://www.w3.org/2001/XMLSchema-instance"
xmlns="urn:hl7-org:v3" xsi:schemaLocation="urn:hl7-org:v3 CDA.xsd">
  <component>
    <structuredBody>
      <component>
        <section>
          <entry>
            <observation classCode="OBS" moodCode="EVN">
              <templateId root="2.16.840.1.113883.10.20.20.2.1"
assigningAuthorityName="GTR Clinical Genomic Statement"/>
              <templateId root="2.16.840.1.113883.10.20.20.3.2"
assigningAuthorityName="GTR Genomic Source Class"/>
              <code/>
              <value xsi:type="CD"/>
              <methodCode/>
            </observation>
          </entryRelationship>
          <entry>
            <observation classCode="OBS" moodCode="EVN">
              <templateId root="2.16.840.1.113883.10.20.20.2.3.4"
assigningAuthorityName="GTR Clinical Genomic Statement Genetic Variation DNA Region Name"/>
              <code code="MTHU008863" codeSystem="2.16.840.1.113883.6.1" codeSystemName="LOINC" displayName="Indications description"/>
              <value xsi:type="CD"/>
            </observation>
          </entryRelationship>
          <entry>
            <observation classCode="OBS" moodCode="EVN">
              <templateId root="2.16.840.1.113883.10.20.20.2.5"
assigningAuthorityName="GTR Interpretive Phenotype Observation"/>
            </observation>
          </entryRelationship>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```
Clinical Genomic Statement Genetic Variation Zygosity

The ClinicalGenomicStatementGeneticVariationDNAChange template is a sub-template of ClinicalGenomicStatement and is used to carry the DNA change of that genetic variation.

1. SHALL conform to GTR Clinical Genomic Statement template (templateId: 2.16.840.1.113883.10.20.20.2)
2. MAY contain [1..1] code/@code = "53034-5" Allelic State (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-100)
3. MAY contain [0..1] value, which MAY be selected from ValueSet 2.16.840.1.113883.10.20.20.9.1 Allelic State STATIC, where its data type is CD (CONF-GTR-101)
4. SHALL satisfy: GTR ClinicalGenomicStatementGeneticVariationDNAChange (self) SHALL satisfy: If self.code@code=53034-5 (LOINC code for Allelic state), then self.value@code SHALL be drawn from the LOINC Value Set OID 2.16.840.1.113883.10.20.20.9.1 (Allelic State: 53034-5). (CONF-GTR-99)
Genomic Source Class

[Observation: templateId 2.16.840.1.113883.10.20.20.3.2]

The GenomicSourceClass template represents 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.

1. **SHALL** conform to *GTR Clinical Genomic Statement* template (templateId: 2.16.840.1.113883.10.20.20.2)
2. **MAY** contain [1..1] code/@code = "48002-0" *Genomic source class* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-81)
3. **MAY** contain [0..1] value, which **MAY** be selected from ValueSet 2.16.840.1.113883.10.20.20.9.12 *Genomic source class* STATIC, where its data type is CD (CONF-GTR-82)
4. **MAY** satisfy: GTR GenomicSourceClass (self) **SHALL** satisfy: If self.code.code=48002-0 (LOINC code for Genomic source class), then self.value.code **SHALL** be drawn from the LOINC answer list 48002-0. (CONF-GTR-80)
<entryRelationship>
  <observation classCode="OBS" moodCode="EVN">
    <templateId root="2.16.840.1.113883.10.20.20.3.3"
      assigningAuthorityName="GTR Indication Observation"/>
    <code code="MTHU008863" codeSystem="2.16.840.1.113883.6.1"
      codeSystemName="LOINC" displayName="Indications description"/>
    <value xsi:type="CD"/>
  </observation>
</entryRelationship>
<entryRelationship>
  <observation classCode="OBS" moodCode="EVN">
    <templateId root="2.16.840.1.113883.10.20.20.2.5"
      assigningAuthorityName="GTR Interpretive Phenotype Observation"/>
  </observation>
</entryRelationship>
<entryRelationship>
  <observation classCode="OBS" moodCode="EVN">
    <templateId root="2.16.840.1.113883.10.20.20.2.1"
      assigningAuthorityName="GTR Clinical Genomic Statement"/>
    <templateId root="2.16.840.1.113883.10.20.20.3.2"
      assigningAuthorityName="GTR Genomic Source Class"/>
    <code/>
    <value xsi:type="CD"/>
    <methodCode/>
  </observation>
</entryRelationship>
</entry>
</section>
</component>
</structuredBody>
</component>
</ClinicalDocument>

Figure 39: Genomic Source Class example

**Indication Observation**

[Observation: templateId 2.16.840.1.113883.10.20.20.3.3]

The IndicationObservation hangs off the genomic observation in the Clinical Genomic Statement template and represent an indication to performing the genomic observation. It can also reside in the IndicationSection and then be referenced from a ClinicalGenomicStatement.

1. **SHALL** conform to CDA Observation
2. **SHALL** contain [1..1] code/@code = "MTHU008863" *Indications description* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-83)
3. **SHALL** contain [1..1] value (CodeSystem: 2.16.840.1.113883.10.20.20.2.1.1 IndicationCode STATIC), where its data type is CD (CONF-GTR-84)
Interpretive Phenotype Observation

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5]

The InterpretivePhenotypeObservation template is an abstract template for sub-templates that represents interpretations of specific types of genomic observation described in the various parts of the GTR.

1. SHALL conform to CDA Observation

<?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">
  <component>
    <structuredBody>
      <component>
        <section>
          <entry>
            <observation classCode="OBS" moodCode="EVN">
              <templateId root="2.16.840.1.113883.10.20.20.2.5"
assigningAuthorityName="GTR Interpretive Phenotype Observation"/>
            </observation>
          </entry>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>

Interpretive Phenotype Observation Cytogenetics

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5.1]

The InterpretivePhenotypeObservationCytogenetics is a sub-template of the InterpretivePhenotypeObservation abstract template, representing interpretations of Cytogenetic testing results.

1. SHALL conform to GTR Interpretive Phenotype Observation template (templateId: 2.16.840.1.113883.10.20.20.2.5)
2. Contains [1..1] code
3. Contains [0..1] value, where its data type is CD

<?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">
  <component>
    <structuredBody>
      <component>
        <section>
          <entry>
            <observation classCode="OBS" moodCode="EVN">
              <templateId root="2.16.840.1.113883.10.20.20.2.5"
assigningAuthorityName="GTR Interpretive Phenotype Observation"/>
            </observation>
          </entry>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
Interpretive Phenotype Observation Gene Expression

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5.2]

The InterpretivePhenotypeObservationGeneExpression is a sub-template of the InterpretivePhenotypeObservation abstract template, representing interpretations of gene expression testing results.

1. SHALL conform to *GTR Interpretive Phenotype Observation* template (templateId: 2.16.840.1.113883.10.20.20.2.5)

2. Contains [1..1] code

3. Contains [0..1] value, where its data type is CD

```xml
<?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">
  <component>
    <structuredBody>
      <component>
        <section>
          <entry>
            <observation classCode="OBS" moodCode="EVN">
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              <templateId root="2.16.840.1.113883.10.20.20.2.5.2" assigningAuthorityName="GTR Interpretive Phenotype Observation Gene Expression"/>
              <code/>
              <value xsi:type="CD"/>
            </observation>
          </entry>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

Figure 43: Interpretive Phenotype Observation Gene Expression example

Interpretive Phenotype Observation Genetic Variation

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5.3]

The InterpretivePhenotypeObservationGeneticVariation is a sub-template of the InterpretivePhenotypeObservation abstract template, representing interpretations of genetic variation testing results.

1. SHALL conform to *GTR Interpretive Phenotype Observation* template (templateId: 2.16.840.1.113883.10.20.20.2.5)
2. MAY contain [1..1] code/@code = "53037-8" Genetic disease sequence variation interpretation (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-52)

3. MAY contain [0..1] value, which MAY be selected from ValueSet 2.16.840.1.113883.10.20.20.9.11 Genetic disease sequence variation interpretation STATIC, where its data type is CD (CONF-GTR-53)

4. MAY satisfy: GTR InterpretivePhenotypeObservationGeneticVariation (self) SHALL satisfy: If self.code.code=53037-8 (LOINC code for Genetic disease sequence variation interpretation), then self.value.code SHALL be drawn from the following LOINC answer list: LA6668-3=Pathogenic; LA6669-1=Presumed pathogenic; LA6682-4=Unknown significance; LA6675-8=Benign; LA6674-1=Presumed benign (CONF-GTR-51)

<?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"><component><structuredBody><component><section><entry><observation classCode="OBS" moodCode="EVN">
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 assigningAuthorityName="GTR Interpretive Phenotype Observation Genetic Variation"/>
 <code/>
 <value xsi:type="CD"/>
 </observation></entry></section></component></structuredBody></component></ClinicalDocument>

Figure 44: Interpretive Phenotype Observation Genetic Variation example

Interpretive Phenotype Observation Pharmacogenomic

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5.4]

The InterpretivePhenotypeObservationPharmacogenomic is a sub-template of the InterpretivePhenotypeObservation abstract template, representing interpretations of pharmacogenomic testing results.

1. SHALL conform to GTR Interpretive Phenotype Observation template (templateId: 2.16.840.1.113883.10.20.20.2.5)

2. MAY contain [1..1] code (CONF-GTR-49)

3. MAY contain [0..1] value, where its data type is CD (CONF-GTR-50)

<?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"><component><structuredBody><component><section><entry><observation classCode="OBS" moodCode="EVN">
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 assigningAuthorityName="GTR Interpretive Phenotype Observation"/>
 <templateId root="2.16.840.1.113883.10.20.20.2.5.4"
 assigningAuthorityName="GTR Interpretive Phenotype Observation Pharmacogenomic"/>
 <code/>
 <value xsi:type="CD"/>
 </observation></entry></section></component></structuredBody></component></ClinicalDocument>
Interpretive Phenotype Observation Pharmacogenomic Drug Efficacy

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5.4.1]

The InterpretivePhenotypeObservationPharmacogenomicDrugEfficacy is a sub-template of the InterpretivePhenotypeObservationPharmacogenomic template, representing interpretations of drug efficacy testing results.

1. **SHALL** conform to *GTR Interpretive Phenotype Observation Pharmacogenomic* template (templateId: 2.16.840.1.113883.10.20.20.2.5.4)

2. **MAY** contain [1..1] code/@code = "51961-1" *Drug efficacy sequence variation interpretation* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-63)

3. **MAY** contain [0..1] value, which **MAY** be selected from ValueSet 2.16.840.1.113883.10.20.20.9.6 *Drug Efficacy Sequence Variation Interpretation* STATIC, where its data type is CD (CONF-GTR-64)

4. **SHALL** satisfy: GTR InterpretivePhenotypeObservationPharmacogenomicDrugEfficacy (self) **SHALL** satisfy: If self.code@code=51961-1 (LOINC code for Drug efficacy sequence variation interpretation), then self.value@code **SHALL** be drawn from the LOINC Value Set 2.16.840.1.113883.10.20.20.9.6 (Drug efficacy sequence variation interpretation: 51961-1). (CONF-GTR-62)
Interpretive Phenotype Observation Pharmacogenomic Drug Metabolism

The InterpretivePhenotypeObservationPharmacogenomicDrugMetabolism is a sub-template of the InterpretivePhenotypeObservationPharmacogenomic template, representing interpretations of drug metabolism testing results.

1. **SHALL** conform to *GTR Interpretive Phenotype Observation Pharmacogenomic* template (templateId: 2.16.840.1.113883.10.20.20.2.5.4)


3. **MAY** contain [0..1] value, which **MAY** be selected from ValueSet 2.16.840.1.113883.10.20.20.9.7 *Drug metabolism sequence variation interpretation* STATIC, where its data type is CD (CONF-GTR-67)

4. **SHALL** satisfy: GTR InterpretivePhenotypeObservationPharmacogenomicDrugMetabolism (self) **SHALL** satisfy: If self.code@code=53040-2 (LOINC code for Drug metabolism sequence variation interpretation), then self.value@code SHALL be drawn from the LOINC Value Set 2.16.840.1.113883.10.20.20.9.7 (Drug metabolism sequence variation interpretation: 53040-2). (CONF-GTR-65)

<?xml version="1.0" encoding="UTF-8"?>

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    <structuredBody>
      <component>
        <section>
          <entry>
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              <templateId root="2.16.840.1.113883.10.20.20.2.5.4" assigningAuthorityName="GTR Interpretive Phenotype Observation Pharmacogenomic"/>
              <templateId root="2.16.840.1.113883.10.20.20.2.5.4.2" assigningAuthorityName="GTR Interpretive Phenotype Observation Pharmacogenomic Drug Metabolism"/>
              <code code="53040-2" codeSystem="2.16.840.1.113883.6.1" codeSystemName="LOINC" display="Drug metabolism sequence variation interpretation"/>
              <value xsi:type="CD"/>
            </observation>
          </entry>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>

---

Overall Interpretive Phenotype Observation Chromosome Analysis

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5.4.2]

1. **SHALL** conform to *GTR Interpretive Phenotype Observation* template (templateId: 2.16.840.1.113883.10.20.20.2.5)

2. **MAY** contain [1..1] code/@code = "62357-9" *Chromosome analysis overall interpretation in Blood or Tissue Qualitative by Molecular genetics method* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26)

3. **MAY** contain [0..1] value, which **MAY** be selected from ValueSet 2.16.840.1.113883.10.20.20.9.3 *Chromosome analysis overall interpretation* STATIC, where its data type is CD
4. **SHALL** satisfy: GTR OverallInterpretivePhenotypeObservationCytogenetics (self) **SHALL** satisfy: If self.code@code=62357-9 (LOINC code for Chromosome analysis overall interpretation in Blood or Tissue Qualitative by Molecular), then self.value@code **SHALL** be drawn from the LOINC Value Set 2.16.840.1.113883.10.20.20.9.3 (Chromosome analysis overall interpretation in Blood or Tissue Qualitative by Molecular: 62357-9).

Figure 48: Overall Interpretive Phenotype Observation Chromosome Analysis example

### Overall Interpretive Phenotype Observation Genetic Disease

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5.5]

The OverallInterpretivePhenotypeObservationGeneticDisease template extends InterpretivePhenotypeObservation and describes the overall interpretation of the genetic variation testing performed.

1. **SHALL** conform to *GTR Interpretive Phenotype Observation* template (templateId: 2.16.840.1.113883.10.20.20.2.5)
2. **MAY** contain [0..1] code/@code = "51968-6" *Genetic Disease Analysis Overall Interpretation* (CodeSystem: 2.16.840.1.113883.6.1 LOINC DYNAMIC 2.26) (CONF-GTR-23)
3. **MAY** contain [0..1] value, which **MAY** be selected from ValueSet 2.16.840.1.113883.10.20.20.9.10 *Genetic disease analysis overall interpretation static*, where its data type is CD (CONF-GTR-24)
4. **SHALL** satisfy: GTR OverallInterpretivePhenotypeObservationGeneticDisease (self) **SHALL** satisfy: If self.code@code=51968-6 (LOINC code for Genetic Disease Analysis Overall Interpretation), then self.value@code **SHALL** be drawn from the LOINC Value Set 2.16.840.1.113883.10.20.20.9.10 (Genetic Disease Analysis Overall Interpretation: 51968-6). (CONF-GTR-22)

```xml
<?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"><component>
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    <section>
      <entry>
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          </templateId>
          <templateId root="2.16.840.1.113883.10.20.20.2.5.5">
            <assigningAuthorityName">GTR Overall Interpretive Phenotype Observation Genetic Disease</assigningAuthorityName>
          </templateId>
          <code/>
          <value xsi:type="CD"/>
        </observation>
      </entry>
    </section>
  </component>
</component>
</ClinicalDocument>
```

Figure 49: Overall Interpretive Phenotype Observation Genetic Disease example

### Overall Interpretive Phenotype Observation Genetic Disease Carrier

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5.6]

The OverallInterpretivePhenotypeObservationGeneticDiseaseCarrier template extends InterpretivePhenotypeObservation and describes the overall interpretation of the genetic disease carrier testing performed.
Overall Interpretive Phenotype Observation Pharmacogenomic Drug Efficacy

The OverallInterpretivePhenotypeObservationPharmacogenomicDrugEfficacy template extends InterpretivePhenotypeObservation and describes the overall interpretation of the pharmacogenomic drug efficacy testing performed.

1. SHALL conform to GTR Interpretive Phenotype Observation template (templateId: 2.16.840.1.113883.10.20.20.2.5)
2. MAY contain [0..1] code/@code = "51964-5" Drug efficacy analysis overall interpretation (CONF-GTR-72)
3. MAY contain [0..1] value, which MAY be selected from ValueSet 2.16.840.1.113883.10.20.20.9.5 Drug efficacy analysis overall interpretation STATIC, where its data type is CD (CONF-GTR-73)
4. SHALL satisfy: GTR OverallInterpretivePhenotypeObservationPharmacogenomicDrugEfficacy (self) SHALL satisfy: If self.code@code=51964-5 (LOINC code for Drug efficacy analysis overall interpretation), then self.value@code SHALL be drawn from the LOINC Value Set 2.16.840.1.113883.10.20.20.9.5 (Drug efficacy analysis overall interpretation: 51964-5). (CONF-GTR-71)
Overall Interpretive Phenotype Observation Pharmacogenomic Drug Metabolism

The OverallInterpretivePhenotypeObservationPharmacogenomicDrugMetabolism template extends InterpretivePhenotypeObservation and describes the overall interpretation of the pharmacogenomic drug Metabolism testing performed.

1. **SHALL** conform to *GTR Interpretive Phenotype Observation Pharmacogenomic* template (templateId: 2.16.840.1.113883.10.20.20.2.5.8)

2. **MAY** contain [1..1] code/@code = "51971-0" *Drug metabolism analysis overall interpretation* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-75)

3. **MAY** contain [0..1] value, which **MAY** be selected from ValueSet 2.16.840.1.113883.10.20.20.9.7 *Drug metabolism analysis overall interpretation STATIC*, where its data type is CD (CONF-GTR-76)

4. **SHALL** satisfy: GTR OverallInterpretivePhenotypeObservationPharmacogenomicDrugMetabolism (self) SHALL satisfy: If self.code/@code=51971-0 (LOINC code for Drug metabolism analysis overall interpretation), then self.value/@code SHALL be drawn from the LOINC Value Set 2.16.840.1.113883.10.20.20.9.7 (Drug metabolism analysis overall interpretation: 51971-0). (CONF-GTR-74)
Test Performed Observation

[Observation: templateId 2.16.840.1.113883.10.20.20.3.4]

The TestPerformedObservation describes one of the tests performed whose results are reported in the GTR. It can be used by the TestsPerformedSection to describe each test in a structured format.

1. SHALL conform to CDA Observation
2. SHOULD contain [0..1] code (CONF-GTR-87)
   - The code attribute should identify the type (or class) of genetic testing at stake, preferably drawn from a published classification / terminology, e.g., "Hereditary Hearing Loss and Deafness" where the value attribute can hold a code representing the test titled "Connexin 26 Full Gene Test", preferably drawn from a published catalog.
3. SHALL contain [1..1] value, where its data type is CD (CONF-GTR-88)
   - The value attribute should be aligned with the semantics of the code assigned to the code attribute, e.g., if code = "Hereditary Hearing Loss and Deafness" then the value attribute can hold a code representing the test titled "Connexin 26 Full Gene Test", preferably drawn from a published catalog.
4. SHOULD contain [0..1] methodCode (CONF-GTR-89)
   - The methodCode can provide more information on the test identified through the value and optionally the code attributes of this observation. The code assigned to methodCode should preferably be drawn from a standard terminology for genetic testing methods.
Chapter 5

OTHER CLASSES

Topics:

- Genetic Specimen
- Performer

This section of the Implementation Guide describes other classes that are not CDA Clinical Documents, Sections, or Clinical Statements.
Genetic Specimen

[Specimen: templateId 2.16.840.1.113883.10.20.20.3.1]

The SpecimenParticipant describes the characteristics of the specimen used in the genetic testing at stake.

1. SHALL conform to CDA Specimen

Figure 54: Genetic Specimen example

Performer

[Performer2: templateId 2.16.840.1.113883.10.20.20.3.2]

The SpecimenParticipant describes the characteristics of the specimen used in the genetic testing at stake.

1. SHALL conform to CDA Performer2

Figure 55: Performer example
Chapter 6

VALUE SETS

Topics:

- Allelic State
- Amino acid change
- Chromosome analysis overall interpretation
- DNA sequence change
- Drug Efficacy Sequence Variation Interpretation
- Drug efficacy analysis overall interpretation
- Drug metabolism analysis overall interpretation
- Drug metabolism sequence variation interpretation
- Genetic disease analysis overall carrier interpretation
- Genetic disease analysis overall interpretation
- Genetic disease sequence variation interpretation
- Genomic source class
- ISCN band level

The following tables summarize the value sets used in this Implementation Guide.
### Allelic State

[OID 2.16.840.1.113883.10.20.20.9.1 from code system: LOINC]

OID: 2.16.840.1.113883.10.20.20.9.1
Name: Allelic State (53034-5)
Code System: 2.16.840.1.113883.6.1
Code System Name: LOINC

<table>
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</thead>
<tbody>
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<td>LOINC</td>
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<tr>
<td>LA6706-1</td>
<td>Heterozygous</td>
<td>2.16.840.1.113883.6.1</td>
<td>LOINC</td>
</tr>
<tr>
<td>LA6703-8</td>
<td>Heteroplasmic</td>
<td>2.16.840.1.113883.6.1</td>
<td>LOINC</td>
</tr>
<tr>
<td>LA6704-6</td>
<td>Homoplasmic</td>
<td>2.16.840.1.113883.6.1</td>
<td>LOINC</td>
</tr>
<tr>
<td>LA6707-9</td>
<td>Hemizygous</td>
<td>2.16.840.1.113883.6.1</td>
<td>LOINC</td>
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</table>

### Amino acid change

[OID 2.16.840.1.113883.10.20.20.9.2 from code system: LOINC]

OID: 2.16.840.1.113883.10.20.20.9.2
Name: Amino acid change type (48006-1)
Code System: 2.16.840.1.113883.6.1
Code System Name: LOINC

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<td>LOINC</td>
</tr>
<tr>
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<td>LOINC</td>
</tr>
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<td>LOINC</td>
</tr>
<tr>
<td>LA9659-9</td>
<td>Insertion and Deletion</td>
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<tr>
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<td>Nonsense</td>
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<td>Silent</td>
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<td>LOINC</td>
</tr>
<tr>
<td>LA6701-2</td>
<td>Stop Codon Mutation</td>
<td>2.16.840.1.113883.6.1</td>
<td>LOINC</td>
</tr>
</tbody>
</table>
Chromosome analysis overall interpretation

OID: 2.16.840.1.113883.10.20.20.9.3 from code system: LOINC

Name: Chromosome analysis overall interpretation in Blood or Tissue Qualitative by Molecular genetics method (62357-9)

Code System: 2.16.840.1.113883.6.1

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<td>Clinical significance unknown</td>
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DNA sequence change

OID: 2.16.840.1.113883.10.20.20.9.4 from code system: LOINC

Name: DNA sequence variation type (48019-4)

Code System: 2.16.840.1.113883.6.1

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<td>Wild type</td>
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<td>LOINC</td>
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<tr>
<td>LA6692-3</td>
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<td>LOINC</td>
</tr>
<tr>
<td>LA6686-5</td>
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Drug Efficacy Sequence Variation Interpretation

OID: 2.16.840.1.113883.10.20.20.9.6 from code system: LOINC

Name: Drug Efficacy Sequence Variation Interpretation (51961-1)

Code System: 2.16.840.1.113883.6.1

Code System Name: LOINC
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</table>

**Drug efficacy analysis overall interpretation**

[OID 2.16.840.1.113883.10.20.20.9.5 from code system: LOINC]

OID: 2.16.840.1.113883.10.20.20.9.5  
Name: Drug efficacy analysis overall interpretation (51964-5)  
Code System: 2.16.840.1.113883.6.1  
Code System Name: LOINC

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**Drug metabolism analysis overall interpretation**

[OID 2.16.840.1.113883.10.20.20.9.7]

OID: 2.16.840.1.113883.10.20.20.9.7  
Name: Drug metabolism analysis overall interpretation (51971-0)

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### Drug metabolism sequence variation interpretation

[OID 2.16.840.1.113883.10.20.20.9.8]

OID: 2.16.840.1.113883.10.20.20.9.8

Name: Drug metabolism sequence variation interpretation (53040-2)

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### Genetic disease analysis overall carrier interpretation

[OID 2.16.840.1.113883.10.20.20.9.9 from code system: LOINC]

OID: 2.16.840.1.113883.10.20.20.9.9

Name: Genetic disease analysis overall carrier interpretation (53039-4)

Code System: 2.16.840.1.113883.6.1

Code System Name: LOINC

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### Genetic disease analysis overall interpretation

[OID 2.16.840.1.113883.10.20.20.9.10 from code system: LOINC]

OID: 2.16.840.1.113883.10.20.20.9.10

Name: Genetic disease analysis overall interpretation (51968-6)

Code System: 2.16.840.1.113883.6.1

Code System Name: LOINC

Defines concept codes for LOINC answer list.

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### Genetic disease sequence variation interpretation

[OID 2.16.840.1.113883.10.20.20.9.11 from code system: LOINC]

**OID:** 2.16.840.1.113883.10.20.9.11

**Name:** Genetic disease sequence variation interpretation (53037-8)

**Code System:** 2.16.840.1.113883.6.1

**Code System Name:** LOINC

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### Genomic source class

[OID 2.16.840.1.113883.10.20.20.9.12 from code system: LOINC]

**OID:** 2.16.840.1.113883.10.20.9.12

**Name:** Genomic source class [Type] in Blood or Tissue by Molecular genetics method (48002-0)

**Code System:** 2.16.840.1.113883.6.1

**Code System Name:** LOINC

The genomic class of the specimen being analyzed: Germline for inherited genome, somatic for cancer genome, and prenatal for fetal genome.

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### ISC N band level

[OID 2.16.840.1.113883.10.20.20.9.13 from code system: LOINC]

**OID:** 2.16.840.1.113883.10.20.9.13

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Name: ISCN band level in Blood or Tissue by Molecular genetics (62358-7)

Code System: 2.16.840.1.113883.6.1

Code System Name: LOINC

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REFERENCES

• LOINC®: Logical Observation Identifiers Names and Codes, Regenstrief Institute.
• SNOMED CT®: SNOMED Clinical Terms SNOMED International Organization.
• Extensible Markup Language, www.w3.org/XML.
• HL7 Clinical Genomics v3 specification. Available through HL7
• HL7 Clinical Genomics v2 specification for genetic testing results message. Available through HL7