

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



**Draft Standard for Trial Use
Second Ballot
May 2011**

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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.

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Revision History

Rev	Date	By Whom	Changes
New	8 January 2009	Amnon Shabo	New
Draft 1	3 August 2010	Amnon Shabo	CGWG review
DSTU R1_O1	10 August 2010	Amnon Shabo	CGWG review
DSTU R1_O2	March 2011	Amnon Shabo	CGWG review

Chapter 1

INTRODUCTION

Topics:

- *Purpose*
- *Approach*
- *Scope*
- *Audience*
- *Organization of This Guide*
- *Use of Templates*
- *Conventions Used in This Guide*
- *Contents of the DSTU Ballot Package*

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 clinicians, we see existing report formats having emphasis on detailed but easy-to-understand interpretations of the testing results along with recommendations. These interpretations may originate from the laboratory or they may be created by a clinician specializing in genetic/genomic medicine. This work also supports, communication 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.

- Convey findings and 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. For XML figures (snippets) are not populated with sample values, which can be found in the complete XML samples enclosed in the ballot package. The template examples in the guide are more of skeletal nature.
2. Constraints on the value attribute have been described in free text similar to Object Constraining Language (OCL) statements. In later releases 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 CDA 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

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`).

Note on DSTU

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).

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).
2. A problem observation SHALL include exactly one Observation / statusCode. (CONF: 815).
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 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.

```
<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

Filename	Description
CDAR2_IG_GENTESTRPT_R1_O2_2011MAY.pdf	This guide
CDA-GeneticTestingReport-GeneticVariation-Sample-v7.xml	A Genetic Testing Report sample with Genetic Variation Sections
CDA-GeneticTestingReport-Cytogenetics-Sample-v1.xml	A Genetic Testing Report sample with a Cytogenetics Section

Filename	Description
cda.xsl	A generic stylesheet for displaying the content of the sample document in HTML

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

[ClinicalDocument: templateId 2.16.840.1.113883.10.20.20]

The GeneticTestingReport is a document template and thus serves as the root template for the GTR Implementation Guide. Its organization is described in the Approach section of this document. The sub-sections residing here constitute the backbone of the GTR. 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)
10. **SHALL** satisfy: Sections and subsections **SHALL** have a title and the title **SHALL NOT** be empty.
11. **SHALL** satisfy: All sections **MAY** occur in any order.

```
<?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>
```

```

    <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>
    <component>
      <section>
        <templateId root="2.16.840.1.113883.10.20.20.1.8"
assigningAuthorityName="GTR Test Details Section"/>
        <templateId root="2.16.840.1.113883.10.20.20.1.2"
assigningAuthorityName="GTR Genetic Variations Section"/>
        <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
codeSystemName="LOINC" displayName="Genetic Variations Section"/>
        <title>Genetic Variations Section</title>
      </section>
    </component>
    <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 code="TBD" codeSystem="2.16.840.1.113883.6.1"
codeSystemName="LOINC" displayName="Gene Expression Section"/>
        <title>Gene Expression Section</title>
      </section>
    </component>
    <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"/>
        <title>Cytogenetics Section</title>
      </section>
    </component>
    <component>
      <section>
        <templateId root="2.16.840.1.113883.10.20.20.1.8"
assigningAuthorityName="GTR Test Details Section"/>
        <templateId root="2.16.840.1.113883.10.20.20.1.6"
assigningAuthorityName="GTR Other Testing Section"/>
        <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
codeSystemName="LOINC" displayName="Genetic Testing Section"/>
        <title>Genetic Testing Section</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 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>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

Figure 6: Background Section example

Cytogenetics Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.4]

The CytogeneticsSection 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 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"/>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

```

    <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
codeSystemName="LOINC" displayName="Cytogenetics Section"/>
    <title>Cytogenetics 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="2.16.840.1.113883.10.20.20.2.2"
assigningAuthorityName="GTR Clinical Genomic Statement Cytogenetics"/>
        <code/>
        <methodCode/>
      </observation>
    </entry>
  </section>
</component>
</structuredBody>
</component>
</ClinicalDocument>

```

Figure 7: Cytogenetics Section example

Findings Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.12]

The FindingSection is a narrative-only section. It resides within the TestDetailsSection and describe in narrative 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.

- SHALL** conform to CDA Section
- 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)
- SHALL** contain [1..1] title = "Findings" (CONF-GTR-48)

```

<?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="2.16.840.1.113883.10.20.20.1.12"
assigningAuthorityName="GTR Findings Section"/>
          <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
codeSystemName="LOINC" displayName="Genetic Testing Findings Section"/>
          <title>Genetic Testing Findings Section</title>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>

```

Figure 8: Findings Section example

Follow Up Genetic Tests Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.1.3]

The FollowUpGenetic TestsSection section resides within the RecommendationSection (within SummarySection) and consists of recommendations for follow-up testing. 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.

- 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 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
2. **SHALL** contain [1..1] code/@code = "TBD" *Genetic Testing Follow Up Visits to Specialists* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-39)
3. **SHALL** contain [1..1] title = "Follow Up Visits To Specialists" (CONF-GTR-40)

```
<?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"
  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="2.16.840.1.113883.10.20.20.1.3"
            assigningAuthorityName="GTR Gene Expression Section"/>
          <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
            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"
  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="2.16.840.1.113883.10.20.20.1.2"
assigningAuthorityName="GTR Genetic Variations Section"/>
        <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
codeSystemName="LOINC" displayName="Genetic Variations Section"/>
        <title>Genetic Variations 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/>
            <methodCode/>
          </observation>
        </entry>
      </section>
    </component>
  </structuredBody>
</component>
</ClinicalDocument>

```

Figure 12: Genetic Variations Section example

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. Common indications for performing a genetic test often include family history of a familial (or inherited) disease or increased risk of developing a disease. Increasingly, molecular analysis of a patient's tumor is another common indication for genetic testing. The IndicationSection 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)

```

<?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="2.16.840.1.113883.10.20.20.1.11"
assigningAuthorityName="GTR Indications Section"/>
          <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
codeSystemName="LOINC" displayName="Genetic Testing Indications Section"/>
          <title>Genetic Testing Indications Section</title>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>

```

```
</ClinicalDocument>
```

Figure 13: Indications Section example

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 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 nests within the TestInformationSection and its text attribute consists of narrative describing methodology of the genetic test at stake. Where possible, use section entries to codify methods used in this genetic tests. Note that LOINC codes are being developed to capture methodology details for sequencing and gene chip tests and might be available around mid 2011.

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 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"/>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

```

        <title>Genetic Testing Methodology</title>
      </section>
    </component>
  </structuredBody>
</component>
</ClinicalDocument>

```

Figure 15: Methodology Section example

Other Testing Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.6]

A 'catcher' section of genomic findings 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)
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-27)
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)

```

<?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="2.16.840.1.113883.10.20.20.1.6"
            assigningAuthorityName="GTR Other Testing Section"/>
          <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
            codeSystemName="LOINC" displayName="Genetic Testing Section"/>
          <title>Genetic Testing 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"/>
              <code/>
              <methodCode/>
            </observation>
          </entry>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>

```

Figure 16: Other Testing Section example

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 tests 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. **SHALL** contain [0..1] `entry`, such that it
 - a. has `@typeCode="COMP" COMP (component)`
 - b. contains *GTR Overall Interpretive Phenotype Chromosome Analysis* (templateId: 2.16.840.1.113883.10.20.20.2.5.4.2)
5. **SHALL** contain [0..1] `entry`, such that it
 - a. has `@typeCode="COMP" COMP (component)`
 - b. contains *GTR Overall Interpretive Phenotype Genetic Disease* (templateId: 2.16.840.1.113883.10.20.20.2.5.5)
6. **SHALL** contain [0..1] `entry`, such that it
 - a. has `@typeCode="COMP" COMP (component)`
 - b. contains *GTR Overall Interpretive Phenotype Genetic Disease Carrier* (templateId: 2.16.840.1.113883.10.20.20.2.5.6)
7. **SHALL** contain [0..1] `entry`, such that it
 - a. has `@typeCode="COMP" COMP (component)`
 - b. contains *GTR Overall Interpretive Phenotype Pharmacogenomic Drug Efficacy* (templateId: 2.16.840.1.113883.10.20.20.2.5.7)
8. **SHALL** contain [0..1] `entry`, such that it
 - a. has `@typeCode="COMP" COMP (component)`
 - b. contains *GTR Overall Interpretive Phenotype Pharmacogenomic Drug Metabolism* (templateId: 2.16.840.1.113883.10.20.20.2.5.8)

```
<?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="2.16.840.1.113883.10.20.20.1.1.1"
            assigningAuthorityName="GTR Overall Interpretation Section"/>
          <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
            codeSystemName="LOINC" displayName="Genetic Testing Overall Interpretation
            Section"/>
          <title>Genetic Testing Overall Interpretation Section</title>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

Figure 17: Overall Interpretation Section example

Recommendations Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.1.5]

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

2. **SHALL** contain [1..1] code/@code = "TBD" *Genetic Testing Recommendations* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-85)
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)

```
<?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="2.16.840.1.113883.10.20.20.1.1.5"
            assigningAuthorityName="GTR Recommendations Section"/>
          <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
            codeSystemName="LOINC" displayName="Genetic Testing Recommendations"/>
          <title>Genetic Testing Recommendations</title>
          <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>
          <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>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

Figure 18: Recommendations Section example

Recommended Actions Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.1.5]

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"

Figure 19: Recommended Actions Section example

References Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.9.3]

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 a narrative describing scientific references of the genetic test, and optionally structured entries representing publications identified through common ids like PubMed ids and OMIM ids. For instance, PubMed id's may be provided as references to peer reviewed journal articles. OMIM id's may be provided to OMIM (Online Mendelian Inheritance in Man) records containing curated information (from peer reviewed literature).

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 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 SpecimenSection 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" *Genetic Testing Specimen and Genomic Source Section* (CodeSystem: 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 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 SummarySection 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. **SHOULD** contain [0..1] component, such that it
 - a. contains *GTR Overall Interpretation Section* (templateId: 2.16.840.1.113883.10.20.20.1.1.1)
5. **SHOULD** contain [0..1] component, such that it
 - a. contains *GTR Tests Performed Section* (templateId: 2.16.840.1.113883.10.20.20.1.10)
6. **SHOULD** contain [0..1] component, such that it
 - a. contains *GTR Test Information Section* (templateId: 2.16.840.1.113883.10.20.20.1.9)
7. **MAY** contain [0..1] component, such that it
 - a. contains *GTR Recommendations Section* (templateId: 2.16.840.1.113883.10.20.20.1.1.5)

```

<?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"
            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>
          <component>
            <section>
              <templateId root="2.16.840.1.113883.10.20.20.1.1.1"
                assigningAuthorityName="GTR Overall Interpretation Section"/>
              <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
                codeSystemName="LOINC" displayName="Genetic Testing Overall Interpretation
                Section"/>
              <title>Genetic Testing Overall Interpretation Section</title>
            </section>
          </component>
        </component>
      </section>
    </structuredBody>
  </component>
</ClinicalDocument>

```

```

    <templateId root="2.16.840.1.113883.10.20.20.1.10"
    assigningAuthorityName="GTR Tests Performed Section"/>
    <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
    codeSystemName="LOINC" displayName="Genetic Tests Performed Section"/>
    <title>Genetic Tests Performed Section</title>
  </section>
</component>
<component>
  <section>
    <templateId root="2.16.840.1.113883.10.20.20.1.9"
    assigningAuthorityName="GTR Test Information Section"/>
    <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
    codeSystemName="LOINC" displayName="Genetic Testing Information"/>
    <title>Genetic Testing Information</title>
  </section>
</component>
<component>
  <section>
    <templateId root="2.16.840.1.113883.10.20.20.1.1.5"
    assigningAuthorityName="GTR Recommendations Section"/>
    <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
    codeSystemName="LOINC" displayName="Genetic Testing Recommendations"/>
    <title>Genetic Testing Recommendations</title>
  </section>
</component>
</section>
</component>
</structuredBody>
</component>
</ClinicalDocument>

```

Figure 22: Summary Section example

Test Details Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.8]

The Test Details Section is the blueprint for all specialized sections that appear on the GTR top level such the genetic variation section, cytogenetic section, etc. It consists of those sub-section 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 appear 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: 2.16.840.1.113883.10.20.20.1.7)
5. **SHOULD** contain [0..1] component, such that it
 - a. contains *GTR Indications Section* (templateId: 2.16.840.1.113883.10.20.20.1.11)
6. **SHOULD** contain [0..1] component, such that it
 - a. contains *GTR Tests Performed Section* (templateId: 2.16.840.1.113883.10.20.20.1.10)
7. **SHOULD** contain [0..1] component, such that it
 - a. contains *GTR Findings Section* (templateId: 2.16.840.1.113883.10.20.20.1.12)
8. **SHOULD** contain [0..1] component, such that it
 - a. contains *GTR Interpretation Section* (templateId: 2.16.840.1.113883.10.20.20.1.13)
9. **SHOULD** contain [0..1] component, such that it

- a. contains *GTR Test Information Section* (templateId: 2.16.840.1.113883.10.20.20.1.9)

```

<?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"/>
          <code/>
          <title/>
          <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>
          <component>
            <section>
              <templateId root="2.16.840.1.113883.10.20.20.1.11"
                assigningAuthorityName="GTR Indications Section"/>
              <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
                codeSystemName="LOINC" displayName="Genetic Testing Indications Section"/>
              <title>Genetic Testing Indications Section</title>
            </section>
          </component>
          <component>
            <section>
              <templateId root="2.16.840.1.113883.10.20.20.1.10"
                assigningAuthorityName="GTR Tests Performed Section"/>
              <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
                codeSystemName="LOINC" displayName="Genetic Tests Performed Section"/>
              <title>Genetic Tests Performed Section</title>
            </section>
          </component>
          <component>
            <section>
              <templateId root="2.16.840.1.113883.10.20.20.1.12"
                assigningAuthorityName="GTR Findings Section"/>
              <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
                codeSystemName="LOINC" displayName="Genetic Testing Findings Section"/>
              <title>Genetic Testing Findings Section</title>
            </section>
          </component>
          <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>
          <component>
            <section>
              <templateId root="2.16.840.1.113883.10.20.20.1.9"
                assigningAuthorityName="GTR Test Information Section"/>

```

```

        <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
codeSystemName="LOINC" displayName="Genetic Testing Information"/>
        <title>Genetic Testing Information</title>
    </section>
</component>
</section>
</component>
</structuredBody>
</component>
</ClinicalDocument>

```

Figure 23: Test Details Section example

Test Information Section

[Section: templateId 2.16.840.1.113883.10.20.20.1.9]

The TestInformationSection nests within the TestDetailsSection and its sub-sections consist of narratives describing information on the genetic tests and corresponding structured data.

1. **SHALL** conform to CDA Section
2. **SHALL** contain [1..1] code/@code = "TBD" *Genetic Testing Information* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-45)
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)

```

<?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"
assigningAuthorityName="GTR Test Information Section"/>
                    <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
codeSystemName="LOINC" displayName="Genetic Testing Information"/>
                    <title>Genetic Testing Information</title>
                    <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>
                    </component>
                    <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>
                </section>
            </component>
        </structuredBody>
    </component>
</ClinicalDocument>

```

```

    </component>
    <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>
  </section>
</component>
</structuredBody>
</component>
</ClinicalDocument>

```

Figure 24: Test Information Section example

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 a narrative describing the tests performed including those which did not have any genetic findings (e.g., no mutations identified within the region examined). It can consist of structured entries describing the performed tests.

1. **SHALL** conform to CDA Section
2. **SHALL** contain [1..1] `@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)

```

<?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="2.16.840.1.113883.10.20.20.1.10"
assigningAuthorityName="GTR Tests Performed Section"/>
          <code code="TBD" codeSystem="2.16.840.1.113883.6.1"
codeSystemName="LOINC" displayName="Genetic Tests Performed Section"/>
          <title>Genetic Tests Performed Section</title>
          <entry>
            <observation classCode="OBS" moodCode="EVN">
              <templateId root="2.16.840.1.113883.10.20.20.3.4"
assigningAuthorityName="GTR Test Performed Observation"/>
              <code/>
              <value xsi:type="CD"/>
              <methodCode/>
            </observation>
          </entry>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>

```

Figure 25: Tests Performed Section example

Chapter

4

CLINICAL STATEMENT TEMPLATES

Topics:

- *Clinical Genomic Statement*
- *Clinical Genomic Statement Cytogenetics*
- *Clinical Genomic Statement Gene Expression*
- *Clinical Genomic Statement Genetic Variation*
- *Cytogenetics Associated Observation Cells Analyzed Count*
- *Cytogenetics Associated Observation Cells Count*
- *Cytogenetics Associated Observation Cells Karyotyped Count*
- *Cytogenetics Associated Observation Colonies Count*
- *Cytogenetics Associated Observation ISCN Band Level*
- *Genetic Variation Associated Observation Amino Acid Change*
- *Genetic Variation Associated Observation DNA Change*
- *Genetic Variation Associated Observation DNA Region Name*
- *Genetic Variation Associated Observation Zygoty*
- *Genomic Associated Observation*
- *Genomic Source Class*
- *Indication Observation*
- *Interpretive Phenotype*
- *Interpretive Phenotype Cytogenetics*
- *Interpretive Phenotype Gene Expression*

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 Genetic Variation*
- *Interpretive Phenotype Pharmacogenomic*
- *Interpretive Phenotype Pharmacogenomic Drug Efficacy*
- *Interpretive Phenotype Pharmacogenomic Drug Metabolism*
- *Overall Interpretive Phenotype Chromosome Analysis*
- *Overall Interpretive Phenotype Genetic Disease*
- *Overall Interpretive Phenotype Genetic Disease Carrier*
- *Overall Interpretive Phenotype Pharmacogenomic Drug Efficacy*
- *Overall Interpretive Phenotype Pharmacogenomic Drug Metabolism*
- *Test Performed Observation*

Clinical Genomic Statement

[Observation: templateId 2.16.840.1.113883.10.20.20.2]

The ClinicalGenomicStatement template serves the structured portion of the GTR Implementation Guide. At its core, there is a genetic observation finding, e.g., a genetic variation, which can be associated with an indication for performing this genetic observation, as well as an association with the interpretation of the observation finding. In addition, it is possible to associate a specimen and genomic source class with the core genetic finding as well as performers of the genetic 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 InterpretivePhonotypeObservation. Nevertheless, due to the specificity of the interpretation, sub-templates of this template further constrain it by using InterpretivePhonotypeObservation sub-templates. For example, ClinicalGenomicStatementGeneticVariation is a sub-template of ClinicalGenomicStatement and is associated with InterpretivePhonotypeObservationGeneticVariation which is a sub-template of InterpretivePhonotypeObservation.

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="SPRT" *SPRT (has support)*
 - b. contains *GTR Interpretive Phenotype* (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. **MAY** contain [0..*] 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 InterpretivePhonotypeObservation template and its sub-templates. (CONF-GTR-8)

```
<?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"/>
              <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>
            </observation>
          </entry>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

```

        </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 26: Clinical Genomic Statement example

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. **MAY** contain [0..1] entryRelationship, such that it
 - a. contains *GTR Interpretive Phenotype Cytogenetics* (templateId: 2.16.840.1.113883.10.20.20.2.5.1)
5. **SHOULD** contain [0..1] entryRelationship, such that it
 - a. contains *GTR Cytogenetics Associated Observation Cells Analyzed Count* (templateId: 2.16.840.1.113883.10.20.20.2.2.1)
6. **SHOULD** contain [0..1] entryRelationship, such that it
 - a. contains *GTR Cytogenetics Associated Observation Cells Count* (templateId: 2.16.840.1.113883.10.20.20.2.2.2)
7. **SHOULD** contain [0..1] entryRelationship, such that it
 - a. contains *GTR Cytogenetics Associated Observation Cells Karyotyped Count* (templateId: 2.16.840.1.113883.10.20.20.2.2.3)
8. **SHOULD** contain [0..1] entryRelationship, such that it
 - a. contains *GTR Cytogenetics Associated Observation Colonies Count* (templateId: 2.16.840.1.113883.10.20.20.2.2.4)
9. **SHOULD** contain [0..1] entryRelationship, such that it
 - a. contains *GTR Cytogenetics Associated Observation ISCN Band Level* (templateId: 2.16.840.1.113883.10.20.20.2.2.5)

10. SHALL satisfy: value SHALL be assigned a string composed using the expression syntax of International System for Human Cytogenetics Nomenclature (ISCN).

```
<?xml 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="2.16.840.1.113883.10.20.20.2.1"
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 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. **MAY** contain [0..1] entryRelationship, such that it
 - a. contains *GTR Interpretive Phenotype Gene Expression* (templateId: 2.16.840.1.113883.10.20.20.2.5.2)
5. **MAY** contain [0..1] entryRelationship, such that it
 - a. contains *GTR Interpretive Phenotype Pharmacogenomic Drug Efficacy* (templateId: 2.16.840.1.113883.10.20.20.2.5.4.1)
6. **MAY** contain [0..1] entryRelationship, such that it
 - a. contains *GTR Interpretive Phenotype Pharmacogenomic Drug Metabolism* (templateId: 2.16.840.1.113883.10.20.20.2.5.4.2)

```
<?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="2.16.840.1.113883.10.20.20.2.3"
                assigningAuthorityName="GTR Clinical Genomic Statement Gene Expression"/>
              <code codeSystemName="HUGO Gene Names"/>
              <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="2.16.840.1.113883.10.20.20.2.1"
                    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 28: Clinical Genomic Statement Gene Expression example

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 single standard for any type 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. The constraining of these nesting observations are described in detail in the associations 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 (identified as HGNC with OID = 2.16.840.1.113883.6.281) or other recognized notation of genetic variations (TBD).
3. **SHOULD** contain [0..1] value (CONF-GTR-55)
 - Please refer to the code attribute documentation.
4. **MAY** contain [0..1] entryRelationship, such that it
 - a. has @typeCode="COMP" *COMP (has component)*
 - b. contains *GTR Interpretive Phenotype Genetic Variation* (templateId: 2.16.840.1.113883.10.20.20.2.5.3)
5. **MAY** contain [0..1] entryRelationship, such that it
 - a. has @typeCode="COMP" *COMP (has component)*
 - b. contains *GTR Interpretive Phenotype Pharmacogenomic Drug Efficacy* (templateId: 2.16.840.1.113883.10.20.20.2.5.4.1)
6. **MAY** contain [0..1] entryRelationship, such that it
 - a. has @typeCode="COMP" *COMP (has component)*
 - b. contains *GTR Interpretive Phenotype Pharmacogenomic Drug Metabolism* (templateId: 2.16.840.1.113883.10.20.20.2.5.4.2)
7. **SHOULD** contain [0..1] entryRelationship, such that it
 - a. contains *GTR Genetic Variation Associated Observation Amino Acid Change* (templateId: 2.16.840.1.113883.10.20.20.2.1.1)
8. **SHOULD** contain [0..1] entryRelationship, such that it
 - a. contains *GTR Genetic Variation Associated Observation DNA Change* (templateId: 2.16.840.1.113883.10.20.20.2.1.2)
9. **SHOULD** contain [0..1] entryRelationship, such that it
 - a. contains *GTR Genetic Variation Associated Observation DNA Region Name* (templateId: 2.16.840.1.113883.10.20.20.2.1.3)

10. SHOULD contain [0..1] entryRelationship, such that it

- a. contains *GTR Genetic Variation Associated Observation Zygosity* (templateId: 2.16.840.1.113883.10.20.20.2.1.4)

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)

```
<?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="2.16.840.1.113883.10.20.20.2.3"
assigningAuthorityName="GTR Clinical Genomic Statement Gene Expression"/>
              <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="2.16.840.1.113883.10.20.20.2.1"
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 29: Clinical Genomic Statement Genetic Variation example

Cytogenetics Associated Observation 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 Genomic Associated Observation* template (templateId: 2.16.840.1.113883.10.20.20.4)
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. **MAY** contain [0..1] value, where its data type is INT

Figure 30: Cytogenetics Associated Observation Cells Analyzed Count example

Cytogenetics Associated Observation 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 Genomic Associated Observation* template (templateId: 2.16.840.1.113883.10.20.20.4)
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 31: Cytogenetics Associated Observation Cells Count example

Cytogenetics Associated Observation 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 Genomic Associated Observation* template (templateId: 2.16.840.1.113883.10.20.20.4)
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 32: Cytogenetics Associated Observation Cells Karyotyped Count example

Cytogenetics Associated Observation 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 Genomic Associated Observation* template (templateId: 2.16.840.1.113883.10.20.20.4)
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 33: Cytogenetics Associated Observation Colonies Count example

Cytogenetics Associated Observation ISCN Band Level

[Observation: templateId 2.16.840.1.113883.10.20.20.2.2.5]

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 Genomic Associated Observation* template (templateId: 2.16.840.1.113883.10.20.20.4)
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. **MAY** contain [0..1] value, which **MAY** 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 34: Cytogenetics Associated Observation ISCN Band Level example

Genetic Variation Associated Observation 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 Genomic Associated Observation* template (templateId: 2.16.840.1.113883.10.20.20.4)
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 type* STATIC, where its data type is CD (CONF-GTR-61)
4. **SHALL** satisfy: GTR ClinicalGenomicStatementGeneticVariationAminoAcidChange (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)
5. **SHALL** satisfy: Either "DNA Change" observation or "Amino Acid Change" observation is required but both may be specified.

Figure 35: Genetic Variation Associated Observation Amino Acid Change example

Genetic Variation Associated Observation 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 Genomic Associated Observation* template (templateId: 2.16.840.1.113883.10.20.20.4)
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 variation change type* 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)
5. **SHALL** satisfy: Either "DNA Change" observation or "Amino Acid Change" observation is required but both may be specified.

Figure 36: Genetic Variation Associated Observation DNA Change example

Genetic Variation Associated Observation DNA Region Name

[Observation: templateId 2.16.840.1.113883.10.20.20.2.1.3]

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 Genomic Associated Observation* template (templateId: 2.16.840.1.113883.10.20.20.4)
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

Figure 37: Genetic Variation Associated Observation DNA Region Name example

Genetic Variation Associated Observation Zygosity

[Observation: templateId 2.16.840.1.113883.10.20.20.2.1.4]

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 Genomic Associated Observation* template (templateId: 2.16.840.1.113883.10.20.20.4)
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)
5. **SHALL** satisfy: If GenomicSourceClass.value@code = "Somatic" then this associated observation **SHALL** NOT be populated, else if GenomicSourceClass.value@code = "Germline" or "Prenatal", then this associated observation **SHALL** be populated.

Figure 38: Genetic Variation Associated Observation Zygosity example

Genomic Associated Observation

[Observation: templateId 2.16.840.1.113883.10.20.20.4]

1. **SHALL** conform to CDA Observation
2. **MAY** contain [1..1] code
3. **MAY** contain [0..1] value
4. **MAY** contain [0..*] methodCode
5. **MAY** contain [0..*] performer, such that it
 - a. contains *GTR Performer* (templateId: 2.16.840.1.113883.10.20.20.3.2)

Figure 39: Genomic Associated Observation example

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)

```
<?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="2.16.840.1.113883.10.20.20.3.2"
                assigningAuthorityName="GTR Genomic Source Class"/>
              <code/>
              <value xsi:type="CD"/>
              <methodCode/>
            </observation>
            <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="2.16.840.1.113883.10.20.20.2.1"
                  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 40: 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 represnet 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)

```
<?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="2.16.840.1.113883.10.20.20.3.3"
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>
          </entry>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

Figure 41: Indication Observation example

Interpretive Phenotype

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5]

The InterpretivePhenotypeObservation template is an abstract template for sub-templates that represent interpretations of specific types of genomic observation described in the various parts of the GTR. It can be associated with performers of the interpretations (note that these performers could be different than those performing the genetic observation itself).

1. **SHALL** conform to CDA Observation
2. **SHOULD** contain [0..*] performer, such that it
 - a. contains *GTR Performer* (templateId: 2.16.840.1.113883.10.20.20.3.2)
 - The performer(s) of the core genomic observations and findings are not necessarily the performer(s) of the interpretation of the findings.
3. **MAY** contain [1..1] code
4. **MAY** contain [0..1] value

Figure 42: Interpretive Phenotype example

Interpretive Phenotype 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* template (templateId: 2.16.840.1.113883.10.20.20.2.5)
2. **MAY** contain [1..1] code
3. **MAY** contain [0..1] value, where its data type is CD

Figure 43: Interpretive Phenotype Cytogenetics example

Interpretive Phenotype 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* template (templateId: 2.16.840.1.113883.10.20.20.2.5)
2. **MAY** contain [1..1] code
3. Contains [0..1] value, where its data type is CD

Figure 44: Interpretive Phenotype Gene Expression example

Interpretive Phenotype 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* 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 2.16.840.1.113883.10.20.9.11 *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 LOINC Value Set 2.16.840.1.113883.10.20.9.11 (Genetic disease sequence variation interpretation: 53037-8) (CONF-GTR-51)

Figure 45: Interpretive Phenotype Genetic Variation example

Interpretive Phenotype 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* 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)

Figure 46: Interpretive Phenotype Pharmacogenomic example

Interpretive Phenotype 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 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)

Figure 47: Interpretive Phenotype Pharmacogenomic Drug Efficacy example

Interpretive Phenotype Pharmacogenomic Drug Metabolism

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5.4.2]

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

1. **SHALL** conform to *GTR Interpretive Phenotype Pharmacogenomic* template (templateId: 2.16.840.1.113883.10.20.20.2.5.4)
2. **MAY** contain [1..1] code/@code = "53040-2" *Drug metabolism sequence variation interpretation* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-66)
3. **MAY** contain [0..1] value, which **MAY** be selected from ValueSet 2.16.840.1.113883.10.20.20.9.8 *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)

Figure 48: Interpretive Phenotype Pharmacogenomic Drug Metabolism example

Overall Interpretive Phenotype Chromosome Analysis

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5.4.2]

1. **SHALL** conform to *GTR Interpretive Phenotype* 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 49: Overall Interpretive Phenotype Chromosome Analysis example

Overall Interpretive Phenotype 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* 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)

Figure 50: Overall Interpretive Phenotype Genetic Disease example

Overall Interpretive Phenotype 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.

1. **SHALL** conform to *GTR Interpretive Phenotype* template (templateId: 2.16.840.1.113883.10.20.20.2.5)
2. **MAY** contain [0..1] code/@code = "53039-4" *Genetic disease analysis overall carrier interpretation* (CodeSystem: 2.16.840.1.113883.6.1 LOINC STATIC 2.26) (CONF-GTR-78)
3. **MAY** contain [0..1] value, which **MAY** be selected from ValueSet 2.16.840.1.113883.10.20.20.9.9 *Genetic disease analysis overall carrier interpretation* STATIC, where its data type is CD (CONF-GTR-79)
4. **SHALL** satisfy: GTR OverallInterpretivePhenotypeObservationGeneticDiseaseCarrier (self) SHALL satisfy: If self.code@code=53039-4 (LOINC code for Genetic disease analysis overall carrier interpretation), then self.value@code SHALL be drawn from the LOINC Value Set 2.16.840.1.113883.10.20.20.9.9 (Genetic disease analysis overall carrier interpretation: 53039-4). (CONF-GTR-77)

Figure 51: Overall Interpretive Phenotype Genetic Disease Carrier example

Overall Interpretive Phenotype Pharmacogenomic Drug Efficacy

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5.7]

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

1. **SHALL** conform to *GTR Interpretive Phenotype Pharmacogenomic* template (templateId: 2.16.840.1.113883.10.20.20.2.5.4)
2. **MAY** contain [1..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)

Figure 52: Overall Interpretive Phenotype Pharmacogenomic Drug Efficacy example

Overall Interpretive Phenotype Pharmacogenomic Drug Metabolism

[Observation: templateId 2.16.840.1.113883.10.20.20.2.5.8]

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

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

Figure 53: Overall Interpretive Phenotype Pharmacogenomic Drug Metabolism example

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.

```
<?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="2.16.840.1.113883.10.20.20.3.4"
                assigningAuthorityName="GTR Test Performed Observation"/>
              <code/>
              <value xsi:type="CD"/>
              <methodCode/>
            </observation>
          </entry>
        </section>
      </component>
    </structuredBody>
  </component>
</ClinicalDocument>
```

Figure 54: Test Performed Observation example

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 charecteristics of the specimen used in the genetic testing at stake.

1. **SHALL** conform to CDA Specimen

Figure 55: Genetic Specimen example

Performer

[Performer2: templateId 2.16.840.1.113883.10.20.20.3.2]

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

1. **SHALL** conform to CDA Performer2

Figure 56: Performer example

Chapter

6

VALUE SETS

Topics:

- *Allelic State*
- *Amino acid change type*
- *Chromosome analysis overall interpretation*
- *DN A sequence variation change type*
- *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*
- *ISC N 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

Code	Display Name	Code System	Code System Name
LA6705-3	Homozygous	2.16.840.1.113883.6.1	LOINC
LA6706-1	Heterozygous	2.16.840.1.113883.6.1	LOINC
LA6703-8	Heteroplasmic	2.16.840.1.113883.6.1	LOINC
LA6704-6	Homoplasmic	2.16.840.1.113883.6.1	LOINC
LA6707-9	Hemizygous	2.16.840.1.113883.6.1	LOINC

Amino acid change type

[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

Code	Display Name	Code System	Code System Name
LA9658-1	Wild type	2.16.840.1.113883.6.1	LOINC
LA6692-3	Deletion	2.16.840.1.113883.6.1	LOINC
LA6686-5	Duplication	2.16.840.1.113883.6.1	LOINC
LA6694-9	Frameshift	2.16.840.1.113883.6.1	LOINC
LA6695-6	Initiating Methionine	2.16.840.1.113883.6.1	LOINC
LA6687-3	Insertion	2.16.840.1.113883.6.1	LOINC
LA9659-9	Insertion and Deletion	2.16.840.1.113883.6.1	LOINC
LA6698-0	Missense	2.16.840.1.113883.6.1	LOINC
LA6699-8	Nonsense	2.16.840.1.113883.6.1	LOINC
LA6700-4	Silent	2.16.840.1.113883.6.1	LOINC
LA6701-2	Stop Codon Mutation	2.16.840.1.113883.6.1	LOINC

Chromosome analysis overall interpretation

[OID 2.16.840.1.113883.10.20.20.9.3 from code system: LOINC]

OID: 2.16.840.1.113883.10.20.20.9.3

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

Code System Name: LOINC

Code	Display Name	Code System	Code System Name
Normal	LA6626-1	2.16.840.1.113883.6.1	LOINC
Abnormal	LA12748-2	2.16.840.1.113883.6.1	LOINC
Clinical significance unkown	LA14007-1	2.16.840.1.113883.6.1	LOINC

DN A sequence variation change type

[OID 2.16.840.1.113883.10.20.20.9.4 from code system: LOINC]

OID: 2.16.840.1.113883.10.20.20.9.4

Name: DNA sequence variation change type (48019-4)

Code System: 2.16.840.1.113883.6.1

Code System Name: LOINC

Code	Display Name	Code System	Code System Name
LA9658-1	Wild type	2.16.840.1.113883.6.1	LOINC
LA6692-3	Deletion	2.16.840.1.113883.6.1	LOINC
LA6686-5	Duplication	2.16.840.1.113883.6.1	LOINC
LA6687-3	Insertion	2.16.840.1.113883.6.1	LOINC
LA6688-1	Insertion/Deletion	2.16.840.1.113883.6.1	LOINC
LA6689-9	Inversion	2.16.840.1.113883.6.1	LOINC
LA6690-7	Substitution	2.16.840.1.113883.6.1	LOINC

Drug Efficacy Sequence Variation Interpretation

[OID 2.16.840.1.113883.10.20.20.9.6 from code system: LOINC]

OID: 2.16.840.1.113883.10.20.20.9.6

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

Code System: 2.16.840.1.113883.6.1

Code System Name: LOINC

Code	Display Name	Code System	Code System Name
LA6676-6	Resistant	2.16.840.1.113883.6.1	LOINC
LA6677-4	Responsive	2.16.840.1.113883.6.1	LOINC
LA9660-7	Presumed resistant	2.16.840.1.113883.6.1	LOINC
LA9661-5	Presumed responsive	2.16.840.1.113883.6.1	LOINC
LA6682-4	Unknown Significance	2.16.840.1.113883.6.1	LOINC
LA6675-8	Benign	2.16.840.1.113883.6.1	LOINC
LA6674-1	Presumed Benign	2.16.840.1.113883.6.1	LOINC
LA9662-3	Presumed non- responsive	2.16.840.1.113883.6.1	LOINC

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

Code	Display Name	Code System	Code System Name
LA6677-4	Responsive	2.16.840.1.113883.6.1	LOINC
LA6676-6	Resistant	2.16.840.1.113883.6.1	LOINC
LA6577-6	Negative	2.16.840.1.113883.6.1	LOINC
LA9663-1	Inconclusive	2.16.840.1.113883.6.1	LOINC
LA9664-9	Failure	2.16.840.1.113883.6.1	LOINC

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)

Code	Display Name	Code System	Code System Name
LA10315-2	Ultrarapid metabolizer	2.16.840.1.113883.6.1	LOINC
LA10316-0	Extensive metabolizer	2.16.840.1.113883.6.1	LOINC
LA10317-8	Intermediate metabolizer	2.16.840.1.113883.6.1	LOINC
LA9657-3	Poor metabolizer	2.16.840.1.113883.6.1	LOINC

Code	Display Name	Code System	Code System Name
LA9663-1	Inconclusive	2.16.840.1.113883.6.1	LOINC

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)

Code	Display Name	Code System	Code System Name
LA10315-2	Ultrarapid metabolizer	2.16.840.1.113883.6.1	LOINC
LA10316-0	Extensive metabolizer	2.16.840.1.113883.6.1	LOINC
LA10317-8	Intermediate metabolizer	2.16.840.1.113883.6.1	LOINC
LA9657-3	Poor metabolizer	2.16.840.1.113883.6.1	LOINC
LA6682-4	Unknown Significance		

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

Code	Display Name	Code System	Code System Name
LA10314-5	Carrier	2.16.840.1.113883.6.1	LOINC
LA6577-6	Negative	2.16.840.1.113883.6.1	LOINC
LA9663-1	Inconclusive	2.16.840.1.113883.6.1	LOINC
LA9664-9	Failure	2.16.840.1.113883.6.1	LOINC

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.

Code	Display Name	Code System	Code System Name
LA6576-8	Positive	2.16.840.1.113883.6.1	LOINC
LA6577-6	Negative	2.16.840.1.113883.6.1	LOINC
LA9663-1	Inconclusive	2.16.840.1.113883.6.1	LOINC
LA9664-9	Failure	2.16.840.1.113883.6.1	LOINC

Genetic disease sequence variation interpretation

[OID 2.16.840.1.113883.10.20.20.9.11 from code system: LOINC]

OID: 2.16.840.1.113883.10.20.20.9.11

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

Code System: 2.16.840.1.113883.6.1

Code System Name: LOINC

Code	Display Name	Code System	Code System Name
LA6668-3	Pathogenic	2.16.840.1.113883.6.1	LOINC
LA6669-1	Presumed pathogenic	2.16.840.1.113883.6.1	LOINC
LA6682-4	Unknown significance	2.16.840.1.113883.6.1	LOINC
LA6675-8	Benign	2.16.840.1.113883.6.1	LOINC
LA6674-1	Presumed benign	2.16.840.1.113883.6.1	LOINC

Genomic source class

[OID 2.16.840.1.113883.10.20.20.9.12 from code system: LOINC]

OID: 2.16.840.1.113883.10.20.20.9.12

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.

Code	Display Name	Code System	Code System Name
LA6683-2	Germline	2.16.840.1.113883.6.1	LOINC
LA6684-0	Somatic	2.16.840.1.113883.6.1	LOINC
LA10429-1	Prenatal	2.16.840.1.113883.6.1	LOINC

ISC N band level

[OID 2.16.840.1.113883.10.20.20.9.13 from code system: LOINC]

OID: 2.16.840.1.113883.10.20.20.9.13

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

Code	Display Name	Code System	Code System Name
LA14008-9	400	2.16.840.1.113883.6.1	LOINC
LA14112-9	425	2.16.840.1.113883.6.1	LOINC
LA14113-7	450	2.16.840.1.113883.6.1	LOINC
LA14009-7	500	2.16.840.1.113883.6.1	LOINC
LA14114-5	550	2.16.840.1.113883.6.1	LOINC
LA14115-2	575	2.16.840.1.113883.6.1	LOINC
LA14010-5	600	2.16.840.1.113883.6.1	LOINC
LA14116-0	650	2.16.840.1.113883.6.1	LOINC
LA14011-3	800	2.16.840.1.113883.6.1	LOINC
LA14012-1	850	2.16.840.1.113883.6.1	LOINC

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- [LOINC[®]](#) : Logical Observation Identifiers Names and Codes, Regenstrief Institute.
- [SNOMED CT[®]](#) : SNOMED Clinical Terms SNOMED International Organization.
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- HL7 Clinical Genomics v3 specification. Available through [HL7](#)
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