Introduction to the SDTM Genomics Findings (GF) Domain

Glenn Barnes, Senior Consultant for Clinical Specimen and Data Management, CDISC
Christine Connolly, Senior Project Manager, Standards Development, CDISC
Dr. Erin Muhlbradt, Clinical/Biomedical Information Specialist, NCI/EVS
Jon Neville, Senior Standards Developer, CDISC

THU 24 MAR 2022
11:00AM-12:30PM ET
Today’s Agenda

1. Housekeeping
2. Speaker Introductions
3. Feature Presentation
4. Upcoming Learning Opportunities & Events
Housekeeping
You will remain on mute
Submit questions at any time via the Questions tool on your GTW app
Housekeeping

Audio Issues?

First, close and restart your GoToWebinar App
Second, check your local internet connection strength using the Audio tool
A recording of this webinar and the slides will be available in the **Members Only** section of CDISC website
Today’s Presenters

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Senior Consultant for Clinical Specimen and Data Management
CDISC

Christine Connolly
Senior Project Manager, Standards Development
CDISC

Dr. Erin Muhlbradt
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CDISC
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Genomics

Genomics refers to the structure, function, evolution, mapping, and editing of an organism’s genome.

Genomic data collected as part of clinical research supports both development of quality patient care and improvements in patient outcomes.

Genomic analysis of subject samples continues to become a standard practice and the methodology for generating these data continues to evolve.
Agenda

1. History
2. Genomics Findings (GF)
3. Future Directions
4. How you can be involved!
History
History

- CDISC began modeling genomic data as early as 2005.

- The Pharmacogenomics/Genetics (PGx) team was formed in 2007 to develop standards.

- From development work a provisional implementation guide was published in May 2015.
  - Study Data Tabulation Model Implementation Guide: Pharmacogenomics/Genetics Version 1.0
    - SDTMIG-PGx v1.0

- Supporting controlled terminology was first published in December 2015.

- After publication genomics continued to evolve with:
  - Increased interest in and feedback for standards
  - New use cases for modeling
The SDTMIG Pharmacogenomics/Genetics v1.0 included seven domains.

- Three domains for specimen data
- Two domains for pharmacogenomics/genetics
- Two domains for biomarkers
History

• The PGx team went on a brief hiatus in June 2017 to regroup and reassess priorities.

• Team reconvened in January 2018 and began review of published standards including:
  • Weekly team meetings with a diverse group of stakeholders
  • Meetings with FDA representatives to get feedback and ask questions
  • Development of refined standards with new use cases
  • Consultation with the CDISC Global Governance Group (GGG)

Team later renamed the CDISC Genomics Subteam
Refined Standards for Genomic Data

A single domain, Genomics Findings (GF), published in the SDTMIG v3.4 in 2021

Deprecation of SDTMIG-PGx v1.0 with:

- Provisional PF domain deprecated and superseded by the GF domain
- Biospecimens domains published in the SDTMIG v3.4 as is and pending updates in future versions
- Provisional PG, PB, and SB domains deprecated with re-instantiation considered if valid use cases are found
**Rationale**

A single domain, Genomics Findings (GF), published in the SDTMIG v3.4

- Biospecimens domains represent data for specimen collection, handling, and processing
- Domains are applicable to use cases beyond genomics and are published Therapeutic Area User Guides (TAUGs) for non-genomic specimens
Rationale

A single domain, Genomics Findings (GF), published in the SDTMIG v3.4

- Biomarker domains represent data for molecular biomarkers of interest for a study and association of defined molecular biomarkers with related subject findings
- Biomarkers are not specific to genomics
- Many types of data are used as biomarkers and data are represented in multiple existing domains
- No additional use cases found
Rationale

A single domain, Genomics Findings (GF), published in the SDTMIG v3.4

- PG domain represents methods and supporting information for genomic testing
- Methods and supporting information are covered in separate non-SDTM dataset files
- May also be applicable to use cases beyond genomic testing
- No additional use cases found
Rationale

A single domain, Genomics Findings (GF), published in the SDTMIG v3.4

- GF is based on PF; PF was refined to develop GF
  - Domain renamed and clarified to accurately describe genomic data
  - Use cases expanded to align with evolving science
  - Variables with overlapping concepts and unclear definitions clarified
  - New concepts added
  - Established SDTM variables added
  - Outdated concepts retired

- Maintaining a separate implementation guide for genomics does not add value
Rationale

Renaming PF to Genomics Findings (GF)

- *Pharmacogenomics/Genetics Findings (PF)* name and definition do not accurately describe data represented in the domain

- Pharmacogenomics and pharmacogenetics are use cases for genomic data
  - **Pharmacogenomics** - Science that examines inherited variations in genes that dictate *drug response* and explores the ways such variations can be used to predict whether a person will respond favorably, adversely, or not at all to an investigational product.
  - **Pharmacogenetics** - Study of the way drugs interact with genetic makeup or the study of genetic response to a drug.

The terms above describe use cases and do not describe genomic data. Additionally, genomic data have many use cases beyond drug response.
Genomics Findings (GF)

• Domain Walkthrough
• Terminology Considerations
• Relationship to Pharmacogenomics/Genetics Findings (PF)
Domain Walkthrough

Let's walkthrough Genomics Findings (GF) scope, record structure, and variables.

We will refer to SDTMIG v3.4 GF Example 2 in this walkthrough.

GF Ex 2 - Single Nucleotide Variation

Created by Dana Booth, last modified on Oct 19, 2021

This example shows findings from an assessment of a known single nucleotide variant in gene ABCG2 using wet laboratory methodology real-time polymerase chain reaction. Findings from this assessment show the genotypes from DNA extracted from the blood of 3 individuals, each with a different genotype at the genetic locus of interest. Because the DNA specimen was extracted from normal blood, the inheritability of the variation is considered to be in the germline.

Row 1: Shows a subject genotype which is homozygous for the variant nucleotide in the reference sequence.
Row 2: Shows a subject genotype which is heterozygous for the nucleotide in the reference sequence.
Row 3: Shows a subject genotype which is homozygous for the nucleotide in the reference sequence.

<table>
<thead>
<tr>
<th>Row</th>
<th>STUDYID</th>
<th>DOMAIN</th>
<th>USUBJID</th>
<th>GFSEQ</th>
<th>GFREFID</th>
<th>GFTESTCD</th>
<th>GFTEST</th>
<th>GFTSTDTL</th>
<th>GFORRES</th>
<th>GFORREF</th>
<th>GFRSTESC</th>
<th>GFRSTREFC</th>
<th>GFINHERT</th>
<th>GFGENREF</th>
<th>GFCHROM</th>
<th>GFSYM</th>
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<tbody>
<tr>
<td>1</td>
<td>C12345</td>
<td>GF</td>
<td>C12345-001</td>
<td>1</td>
<td>NA18537</td>
<td>SNV</td>
<td>Single Nucleotide Variation</td>
<td>GENOTYPE</td>
<td>T/T</td>
<td>G/G</td>
<td>T/T</td>
<td>G/G</td>
<td>GERMLINE VARIATION</td>
<td>GRCh38:p13</td>
<td>4</td>
<td>ABCG2</td>
</tr>
</tbody>
</table>
GF Domain Scope

**GF Ex 2 - Single Nucleotide Variation**

This example shows findings from an assessment of a known single nucleotide variant in gene ABCG2 using wet laboratory methodology real-time polymerase chain reaction. Findings from this assessment show the genotypes from DNA extracted from the blood of 3 individuals, each with a different genotype at the genetic locus of interest. Because the DNA specimen was extracted from normal blood, the inheritability of the variation is considered to be in the germline.

Representation of findings related to the structure, function, evolution, mapping, and editing of subject and non-host organism genomic material of interest; i.e.,:

- Genetic variation
- Transcription
- Summary measures derived from these assessments

Such findings include inferences/predictions about related proteins/amino acids

- However, direct assessments of proteins (e.g., of amino acids) are out of scope for this domain.
GF Domain Scope

For non-host organisms including bacteria, viruses, and parasites:

• Genetic findings from assessments of non-host organisms in subject samples are in scope for GF

• The following are not in scope; findings for:
  • Detection or determination of the identity of a viable, non-host organism or infectious agent (Microbiology Specimen (MB) domain)
  • Determination of the resistance/susceptibility of a non-host organism to a drug (Microbiology Susceptibility (MS) domain)
GF Ex 2 - Single Nucleotide Variation

This example shows findings from an assessment of a known single nucleotide variant in gene ABCG2 using wet laboratory methodology real-time polymerase chain reaction. Findings from this assessment show the genotypes from DNA extracted from the blood of 3 individuals, each with a different genotype at the genetic locus of interest. Because the DNA specimen was extracted from normal blood, the inheritability of the variation is considered to be in the germline.

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<th>GFORRES</th>
<th>GFORREF</th>
<th>GFSTRES</th>
<th>GFSTREFC</th>
<th>GFINHERN</th>
<th>GFGENREF</th>
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<td>GERMLINE VARIATION</td>
<td>GRCh38.p13</td>
<td>4</td>
<td>ABCG2</td>
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</table>

Expected structure is one record per finding per observation per biospecimen per subject
GF Variables

GF Ex 2 - Single Nucleotide Variation

Created by Dana Booth, last modified on Oct 19, 2021

This example shows findings from an assessment of a known single nucleotide variant in gene ABCG2 using wet laboratory methodology real-time polymerase chain reaction. Findings from this assessment show the genotypes from DNA extracted from the blood of 3 individuals, each with a different genotype at the genetic locus of interest. Because the DNA specimen was extracted from normal blood, the inheritability of the variation is considered to be in the germline.

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GF is comprised of fifty-seven variables; 11 Identifiers, 1 Topic, 35 Qualifiers, 10 Timing
**GF Identifier Variables**

**GF Ex 2 - Single Nucleotide Variation**

<table>
<thead>
<tr>
<th>Variable Name</th>
<th>Variable Label</th>
<th>Type</th>
<th>Controlled Terms, Codelist or Format</th>
<th>Role</th>
<th>CDISC Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>STUDYID</td>
<td>Study Identifier</td>
<td>Char</td>
<td></td>
<td>Identifier</td>
<td>Unique identifier for a study.</td>
</tr>
<tr>
<td>DOMAIN</td>
<td>Domain Abbreviation</td>
<td>Char</td>
<td>GF</td>
<td>Identifier</td>
<td>Two-character abbreviation for the domain.</td>
</tr>
<tr>
<td>USUBJID</td>
<td>Unique Subject Identifier</td>
<td>Char</td>
<td></td>
<td>Identifier</td>
<td>Identifier used to uniquely identify a subject across all studies for all applications or submissions involving the product.</td>
</tr>
<tr>
<td>SPDEVID</td>
<td>Sponsor Device Identifier</td>
<td>Char</td>
<td></td>
<td>Identifier</td>
<td>Sponsor-defined identifier for a device.</td>
</tr>
<tr>
<td>NHOID</td>
<td>Non-Host Organism Identifier</td>
<td>Char</td>
<td></td>
<td>Identifier</td>
<td>Sponsor-defined identifier for a non-host organism which should only be used when the organism is the subject of the TEST. This variable should be populated with an intuitive name based on the identity of the non-host organism as reported by a lab (e.g., &quot;A/California/7/2009 (H1N1)&quot;), it is not to be used as a qualifier of the result in the record on which it appears.</td>
</tr>
<tr>
<td>GFSEQ</td>
<td>Sequence Number</td>
<td>Num</td>
<td></td>
<td>Identifier</td>
<td>Sequence number to ensure uniqueness of records within a dataset for a subject. May be any valid number (including decimals) and does not have to start at 1.</td>
</tr>
<tr>
<td>GFGRPID</td>
<td>Group ID</td>
<td>Char</td>
<td></td>
<td>Identifier</td>
<td>Used to link together a block of related records within a subject in a domain.</td>
</tr>
<tr>
<td>GREFID</td>
<td>Reference ID</td>
<td>Char</td>
<td></td>
<td>Identifier</td>
<td>A unique identifier for the assayed genetic specimen.</td>
</tr>
<tr>
<td>GFSPID</td>
<td>Sponsor-Defined Identifier</td>
<td>Char</td>
<td></td>
<td>Identifier</td>
<td>Sponsor-defined identifier.</td>
</tr>
<tr>
<td>GFLNKID</td>
<td>Link ID</td>
<td>Char</td>
<td></td>
<td>Identifier</td>
<td>Identifier used to link related records across domains. This may be a one-to-one or a one-to-many relationship.</td>
</tr>
<tr>
<td>GFLNKGRP</td>
<td>Link Group ID</td>
<td>Char</td>
<td></td>
<td>Identifier</td>
<td>Identifier used to link related records across domains. This will usually be a many-to-one relationship.</td>
</tr>
</tbody>
</table>

Platform used to detect the finding may be represented here.
### GF Topic Variable

**GF Ex 2 - Single Nucleotide Variation**

<table>
<thead>
<tr>
<th>Variable Name</th>
<th>Variable Label</th>
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<th>Role</th>
<th>CDISC Notes</th>
<th>Core</th>
</tr>
</thead>
<tbody>
<tr>
<td>GFTESTCD</td>
<td>Short Name of Genomic Measurement</td>
<td>Char</td>
<td>(GFTESTCD)</td>
<td>Topic</td>
<td>Short name of the measurement, test, or examination described in GFTEST. It can be used as a column name when converting a dataset from a vertical to a horizontal format. The value in GFTESTCD cannot be longer than 8 characters, nor can it start with a number (e.g., “TTEST” is not valid). GFTESTCD cannot contain characters other than letters, numbers, or underscores.</td>
<td>Req</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>STUDYID</th>
<th>DOMAIN</th>
<th>USUBJID</th>
<th>GFSEQ</th>
<th>GFREFID</th>
<th>GFTESTCD</th>
<th>GFTEST</th>
<th>GFTSTDTL</th>
<th>GFORRES</th>
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<tr>
<td>C12345</td>
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<td>SNV</td>
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</tr>
</tbody>
</table>
# GF Qualifier Variables

## GF Ex 2 - Single Nucleotide Variation

<table>
<thead>
<tr>
<th>Variable Name</th>
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<th>Role</th>
<th>CDISC Notes</th>
<th>Core</th>
</tr>
</thead>
<tbody>
<tr>
<td>GFTEST</td>
<td>Name of Genomic Measurement</td>
<td>Char</td>
<td>(GFTEST)</td>
<td>Synonym Qualifier</td>
<td>Long name for GFTESTCD. The value in GFTEST cannot be longer than 40 characters.</td>
<td>Req</td>
</tr>
<tr>
<td>GFTSTDTL</td>
<td>Measurement, Test, or Examination Detail</td>
<td>Char</td>
<td>(GFTSTDTL)</td>
<td>Variable Qualifier</td>
<td>Description of a reportable qualifying the assessment in GFTESTCD and GFTEST.</td>
<td>Perm</td>
</tr>
<tr>
<td>GFCAT</td>
<td>Category for Genomic Finding</td>
<td>Char</td>
<td></td>
<td>Grouping Qualifier</td>
<td>Used to define a category of topic-variable values.</td>
<td>Perm</td>
</tr>
<tr>
<td>GFSCAT</td>
<td>Subcategory for Genomic Finding</td>
<td>Char</td>
<td></td>
<td>Grouping Qualifier</td>
<td>Used to define a further categorization of GFCAT values.</td>
<td>Perm</td>
</tr>
</tbody>
</table>
### GF Qualifier Variables

<table>
<thead>
<tr>
<th>Variable Name</th>
<th>Variable Label</th>
<th>Type</th>
<th>Role</th>
<th>CDISC Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>GFORRES</td>
<td>Result or Finding in Original Units</td>
<td>Char</td>
<td>Result Qualifier</td>
<td>Result of the measurement or finding as originally received or collected.</td>
</tr>
<tr>
<td>GFORRESU</td>
<td>Original Units</td>
<td>Char</td>
<td>Variable Qualifier</td>
<td>Unit for GFORRES.</td>
</tr>
<tr>
<td>GFORREF</td>
<td>Reference Result in Original Units</td>
<td>Char</td>
<td>Variable Qualifier</td>
<td>Reference value for the result or finding as originally received or collected. GFORREF uses the same units as GFORRES, if applicable.</td>
</tr>
<tr>
<td>GFSTREC</td>
<td>Result or Finding in Standard Format</td>
<td>Char</td>
<td>Result Qualifier</td>
<td>Contains the result value for all findings, copied or derived from GFORRES, in a standard format or in standard units. GFSTREC should store all results or findings in character format; if results are numeric, they should also be stored in numeric format in GFSTRESN.</td>
</tr>
<tr>
<td>GFSTRESN</td>
<td>Numeric Result/Finding in Standard Units</td>
<td>Num</td>
<td>Result Qualifier</td>
<td>Used for continuous or numeric results or findings in standard format; copied in numeric format from GFSTREC. GFSTRESN should store all numeric test results or findings.</td>
</tr>
<tr>
<td>GFSTRESU</td>
<td>Standard Units</td>
<td>Char</td>
<td>Variable Qualifier</td>
<td>Standardized units used for GFSTREC, GFSTRESN, GFSTRECF, and GFSTREFN.</td>
</tr>
<tr>
<td>GFSTREFC</td>
<td>Reference Result in Standard Format</td>
<td>Char</td>
<td>Variable Qualifier</td>
<td>Reference value for the result or finding copied or derived from GFORREF in a standard format.</td>
</tr>
<tr>
<td>GFSTREFN</td>
<td>Numeric Reference Result in Std Units</td>
<td>Num</td>
<td>Variable Qualifier</td>
<td>Reference value for continuous or numeric results or findings in standard format or in standard units. GFSTREFN uses the same units as GFSTRESN, if applicable.</td>
</tr>
<tr>
<td>GFRESCAT</td>
<td>Result Category</td>
<td>Char</td>
<td>Variable Qualifier</td>
<td>Used to categorize the result of a finding.</td>
</tr>
</tbody>
</table>

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**GF Ex 2 - Single Nucleotide Variation**

| GFTESTCD | GFTEST | GFSTDTL | GFORRES | GFORREF | GFSTREC | GFSTRECF | GFINHERT | GFGENREF | GFCHROM | GFSYM | GFSYMTYP | GFGENLOC | GFSEQID | GFPRID | GFNAM | GFSPEC | GFMETHOD |
|----------|--------|---------|---------|---------|---------|----------|----------|----------|---------|-------|---------|----------|----------|--------|-------|--------|--------|----------|
| SNV      |        |         | T/T     | G/G     | T/T     | G/G      |          |          |         |       |         |          |          |        |       |        |        |          |

---

1. [GFORRES] is marked as a critical value in the GF Ex 2 - Single Nucleotide Variation section.
# GF Qualifier Variables

## GF Ex 2 - Single Nucleotide Variation

<table>
<thead>
<tr>
<th>Variable Name</th>
<th>Variable Label</th>
<th>Type</th>
<th>Controlled Terms, Codelist or Format</th>
<th>Role</th>
<th>CDISC Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>GFINHERT</td>
<td>Inheritability</td>
<td>Char</td>
<td>(INHERTGF)</td>
<td>Variable Qualifier</td>
<td>Identifies whether the variation can be passed to the next generation.</td>
</tr>
<tr>
<td>GFGENREF</td>
<td>Genome Reference</td>
<td>Char</td>
<td></td>
<td>Variable Qualifier</td>
<td>An identifier for the genome reference used to generate the reported result. For example, Genome Reference Consortium Human Build 38 patch release 13 may be represented as &quot;GRCh38,p13&quot;.</td>
</tr>
<tr>
<td>GFCHROM</td>
<td>Chromosome Identifier</td>
<td>Char</td>
<td></td>
<td>Variable Qualifier</td>
<td>The designation (name or number) of the chromosome or contig on which the variant or other feature appears (e.g., &quot;17&quot;, &quot;X&quot;).</td>
</tr>
<tr>
<td>GFSYM</td>
<td>Genomic Symbol</td>
<td>Char</td>
<td>*</td>
<td>Variable Qualifier</td>
<td>A published symbol for the portion of the genome serving as a locus for the experiment/test.</td>
</tr>
<tr>
<td>GFSYMTYP</td>
<td>Genomic Symbol Type</td>
<td>Char</td>
<td>(SYMTYPGF)</td>
<td>Variable Qualifier</td>
<td>A description of the type of genomic entity that is represented by the published symbol in GFSYM.</td>
</tr>
<tr>
<td>GFGENLOC</td>
<td>Genetic Location</td>
<td>Char</td>
<td></td>
<td>Variable Qualifier</td>
<td>Specifies the location within a sequence for the observed value in GFORRES.</td>
</tr>
<tr>
<td>GFGENSRC</td>
<td>Genetic Sub-Region</td>
<td>Char</td>
<td></td>
<td>Variable Qualifier</td>
<td>The portion of the locus in which the variation was found. Examples: &quot;Exon 15&quot;, &quot;Kinase domain&quot;.</td>
</tr>
<tr>
<td>GFSSEQID</td>
<td>Sequence Identifier</td>
<td>Char</td>
<td></td>
<td>Variable Qualifier</td>
<td>A unique identifier for the sequence used as the reference to identify the genetic variation in the result. Examples: &quot;NM_001234&quot;, &quot;ENSG00000182333&quot;, &quot;ENST00000343849.2&quot;.</td>
</tr>
<tr>
<td>GFVNRID</td>
<td>Published Variant Identifier</td>
<td>Char</td>
<td></td>
<td>Variable Qualifier</td>
<td>A unique identifier for the variation that has been publicly characterized in an external database. Examples: &quot;rs22331142&quot;, &quot;COSM41596&quot;.</td>
</tr>
<tr>
<td>GFCOPYID</td>
<td>Copy Identifier</td>
<td>Char</td>
<td></td>
<td>Variable Qualifier</td>
<td>An arbitrary identifier used to differentiate between copies of a genetic target of interest present on homologous chromosomes.</td>
</tr>
</tbody>
</table>

**New variables for genomics in SDTM v2.0**
# GF Qualifier Variables

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<tr>
<th>GFTESTCD</th>
<th>GFTEST</th>
<th>GFTSTDNL</th>
<th>GFORRES</th>
<th>GFORREF</th>
<th>GFSTRESC</th>
<th>GFSTREFC</th>
<th>GFPHRT</th>
<th>GFCHROM</th>
<th>GFSYM</th>
<th>GFSYMTYP</th>
<th>GFGENLOC</th>
<th>GFSEQID</th>
<th>GPERID</th>
<th>GFNAME</th>
<th>GFSPIC</th>
<th>GFMETHOD</th>
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<tr>
<td>SNV</td>
<td>Single Nucleotide Variation</td>
<td>GENOTYPE</td>
<td>T/T</td>
<td>G/G</td>
<td>T/T</td>
<td>G/G</td>
<td>GERMLINE VARIATION</td>
<td>GRCH38:123</td>
<td>4</td>
<td>ABCG2</td>
<td>GENE WITH PROTEIN PRODUCT</td>
<td>488131171</td>
<td>ENSG00000118777</td>
<td>rs2231142</td>
<td>AOME LABS</td>
<td>DNA</td>
</tr>
</tbody>
</table>

### Variable Name | Variable Label | Type | Controlled Terms, Codelist or Format | Role | CDISC Notes | Core |
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<thead>
<tr>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>GFSTAT</td>
<td>Completion Status</td>
<td>Char</td>
<td>(ND)</td>
<td>Record Qualifier</td>
<td>Used to indicate that a question was not asked or a test was not done, or a test was attempted but did not generate a result. Should be null or have a value of &quot;NOT DONE&quot;.</td>
<td>Perm</td>
</tr>
<tr>
<td>GFREASND</td>
<td>Reason Test Not Done</td>
<td>Char</td>
<td></td>
<td>Record Qualifier</td>
<td>Reason not done. Used in conjunction with GFSTAT when value is &quot;NOT DONE&quot;.</td>
<td>Perm</td>
</tr>
<tr>
<td>GFXFN</td>
<td>External File Path</td>
<td>Char</td>
<td></td>
<td>Record Qualifier</td>
<td>The filename and/or path to external data not stored in the same format and possibly not the same location as the other data for a study.</td>
<td>Perm</td>
</tr>
<tr>
<td>GFNAM</td>
<td>Laboratory/Vendor Name</td>
<td>Char</td>
<td></td>
<td>Record Qualifier</td>
<td>Name or identifier of the vendor that provided the test result. When more than 1 vendor is involved in the generation of the result, additional vendors should be represented as supplemental qualifiers.</td>
<td>Perm</td>
</tr>
<tr>
<td>GFSPIC</td>
<td>Specimen Material Type</td>
<td>Char</td>
<td>(GENSMP)</td>
<td>Record Qualifier</td>
<td>Identifies the type of genetic material used for the measurement.</td>
<td>Perm</td>
</tr>
</tbody>
</table>
# GF Qualifier Variables

## GF Ex 2 - Single Nucleotide Variation

<table>
<thead>
<tr>
<th>Variable Name</th>
<th>Variable Label</th>
<th>Type</th>
<th>Controlled Terms, Codelist or Format</th>
<th>Role</th>
<th>CDISC Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>GFMETHOD</td>
<td>Method of Test or Examination</td>
<td>Char</td>
<td>(METHOD)</td>
<td>Record Qualifier</td>
<td>The test method by which the examination is performed by the wet lab in order to yield the result reported in the dataset.</td>
</tr>
<tr>
<td>GFRUNID</td>
<td>Run ID</td>
<td>Char</td>
<td></td>
<td>Record Qualifier</td>
<td>A unique identifier for a particular run of a test performed by the wet lab on a particular batch of samples. This identifier can be used to distinguish between records for the same test performed at different times.</td>
</tr>
<tr>
<td>GFANMETH</td>
<td>Analysis Method</td>
<td>Char</td>
<td>(GFANMET)</td>
<td>Record Qualifier</td>
<td>The method of secondary processing performed by the dry lab to yield the result reported in the dataset.</td>
</tr>
<tr>
<td>GFBFLFL</td>
<td>Baseline Flag</td>
<td>Char</td>
<td>(NY)</td>
<td>Record Qualifier</td>
<td>Indicator used to identify a baseline value. Should be “Y” or null.</td>
</tr>
<tr>
<td>GFDRVFL</td>
<td>Derived Flag</td>
<td>Char</td>
<td>(NY)</td>
<td>Record Qualifier</td>
<td>Used to indicate a derived record (e.g., a record that represents the average of other records such as a computed baseline). Should be “Y” or null.</td>
</tr>
<tr>
<td>GFLLOQ</td>
<td>Lower Limit of Quantitation</td>
<td>Num</td>
<td></td>
<td>Variable Qualifier</td>
<td>Indicates the lower limit of quantitation for an assay. Units will be those used for GFSTRESU.</td>
</tr>
<tr>
<td>GFREPNUM</td>
<td>Repetition Number</td>
<td>Num</td>
<td></td>
<td>Record Qualifier</td>
<td>The instance number of a test that is repeated within a given timeframe for the same test performed by the wet lab.</td>
</tr>
</tbody>
</table>

**Core**:

- Exp: Experimental
- Perm: Permitted
# GF Timing Variables

## GF Ex 2 - Single Nucleotide Variation

<table>
<thead>
<tr>
<th>Variable Name</th>
<th>Variable Label</th>
<th>Type</th>
<th>Controlled Terms, Codelist or Format</th>
<th>Role</th>
<th>CDISC Notes</th>
</tr>
</thead>
<tbody>
<tr>
<td>VISITNUM</td>
<td>Visit Number</td>
<td>Num</td>
<td>ISO 8601 date or interval</td>
<td>Timing</td>
<td>Clinical encounter number. Numeric version of VISIT, used for sorting.</td>
</tr>
<tr>
<td>VISIT</td>
<td>Visit Name</td>
<td>Char</td>
<td>ISO 8601 date or interval</td>
<td>Timing</td>
<td>Protocol-defined description of clinical encounter.</td>
</tr>
<tr>
<td>VISITDY</td>
<td>Planned Study Day of Visit</td>
<td>Num</td>
<td>ISO 8601 date or interval</td>
<td>Timing</td>
<td>Planned study day of VISIT. Should be an integer.</td>
</tr>
<tr>
<td>GFDTC</td>
<td>Date/Time of Specimen Collection</td>
<td>Char</td>
<td>ISO 8601 date or interval</td>
<td>Timing</td>
<td>Date and time of specimen collection.</td>
</tr>
<tr>
<td>GFDY</td>
<td>Study Day of Specimen Collection</td>
<td>Num</td>
<td>ISO 8601 date or interval</td>
<td>Timing</td>
<td>Actual study day of visit/collection/exam expressed in integer days relative to the sponsor-defined RFSTDTDC in Demographics.</td>
</tr>
<tr>
<td>GFTPT</td>
<td>Planned Time Point Name</td>
<td>Char</td>
<td>ISO 8601 duration</td>
<td>Timing</td>
<td>Text description of time when a measurement or observation should be taken as defined in the protocol. This may be represented as an elapsed time relative to a fixed reference point, such as time of last dose. See GFTPTNUM and GFTPTREF.</td>
</tr>
<tr>
<td>GFTPTNUM</td>
<td>Planned Time Point Number</td>
<td>Num</td>
<td>ISO 8601 duration</td>
<td>Timing</td>
<td>Numerical version of GFTPT used in sorting.</td>
</tr>
<tr>
<td>GFELTM</td>
<td>Planned Elapsed Time from Time Point Ref</td>
<td>Char</td>
<td>ISO 8601 duration</td>
<td>Timing</td>
<td>Elapsed time relative to a planned fixed reference (GFTPTREF). This variable is useful where there are repetitive measures. Not a clock time or a date time variable, but an interval, represented as ISO duration.</td>
</tr>
<tr>
<td>GFTPTREF</td>
<td>Time Point Reference</td>
<td>Char</td>
<td>ISO 8601 duration</td>
<td>Timing</td>
<td>Name of the fixed reference point referred to by GFELTM, GFTPTNUM, and GFTPT. Examples: &quot;PREVIOUS DOSE&quot;, &quot;PREVIOUS MEAL&quot;.</td>
</tr>
<tr>
<td>GFRFTDTC</td>
<td>Date/Time of Reference Time Point</td>
<td>Char</td>
<td>ISO 8601 date or interval</td>
<td>Timing</td>
<td>Date/time for a fixed reference time point defined by GFTPTREF.</td>
</tr>
</tbody>
</table>

1. ISO 8601 is an international standard for date and time representations.
## Terminology Considerations

### GF variables with Controlled Terminology

<table>
<thead>
<tr>
<th>Variable Name</th>
<th>Variable Label</th>
<th>Description</th>
<th>Associated Controlled Terminology?</th>
</tr>
</thead>
<tbody>
<tr>
<td>GFTEST/CD</td>
<td>Name/Short Name of Genomic Measurement</td>
<td>Long/short name of the measurement, test, or examination described in GFTEST.</td>
<td>CDISC CT</td>
</tr>
<tr>
<td>GFTSTDTL</td>
<td>Measurement, Test, or Examination Detail</td>
<td>Description of a reportable qualifying the assessment in GFTESTCD and GFTEST.</td>
<td>CDISC CT</td>
</tr>
<tr>
<td>GFINHERT</td>
<td>Inheritability</td>
<td>Identifies whether the variation can be passed to the next generation.</td>
<td>CDISC CT</td>
</tr>
<tr>
<td>GFGENREF</td>
<td>Genome Reference</td>
<td>An identifier for the genome reference used to generate the reported result.</td>
<td>External</td>
</tr>
<tr>
<td>GFSYM</td>
<td>Genomic Symbol</td>
<td>A published symbol for the portion of the genome serving as a locus for the experiment/test.</td>
<td>External-HGNC</td>
</tr>
<tr>
<td>GFSYMTYP</td>
<td>Genomic Symbol Type</td>
<td>A description of the type of genomic entity that is represented by the published symbol in --SYM.</td>
<td>CDISC-C T</td>
</tr>
<tr>
<td>GFSEQID</td>
<td>Sequence Identifier</td>
<td>A unique identifier for the sequence used as the reference to identify the genetic variation in the result.</td>
<td>External</td>
</tr>
<tr>
<td>GFPRVID</td>
<td>Published Variant Identifier</td>
<td>A unique identifier for the variation that has been publicly characterized in an external database.</td>
<td>External</td>
</tr>
</tbody>
</table>
Terminology Considerations for GFTEST/CD and GFTSTDTL

• These are closely coupled:
  • The value in GFTEST/CD represents a high level or generalized description of the assessment, which is considered a characteristic finding of the genomic material.
  • The value in GFTSTDTL represents the specific reportable for the assessment described in the GFTEST/CD value.

• A GFTEST/CD value may have one or more associated GFTSTDTL values and a single GFTSTDTL value may be associated with more than one GFTEST/CD value.

• To explicitly describe findings reported in GF, a value for GFTEST/CD and GFTSTDTL are generally both needed.

• When submitting a CDISC Change Request for either codelist, please include both GFTEST/CD and GFTSTDTL values in the request so that the terminology team may better understand the context for the request.
## Terminology Considerations for GFTEST/CD and GFTSTDTTL

### GF Ex 2 - Single Nucleotide Variation

| STUDYID | DOMAIN | USUBJID | SPDEVID | GFSSEQ | GFSRPIP | GFIPID | GTFTESTCD | GFTTEST | GFTSTOTL | GFORRESU | GFSTRES | GFSINST | GFSYMP | GFCHROM | GFSYM | GFSTDTDTL | GFSORB | GFSYMTPY | GFGENLOC |
|---------|--------|---------|---------|--------|----------|--------|------------|----------|----------|----------|---------|---------|---------|---------|---------|-------|-----------|--------|----------|----------|
| ABC-123 | GF     | 123101  | ACME    | 1      | TRF001300 | SNV    | D1853N     | D1853N   | SOMATIC  | GRCH3.75 | 11      | ATM     | GENE WITH PROTEIN PRODUCT |
| ABC-123 | GF     | 123101  | ACME    | 2      | TRF001300 | SNV    | 5557G>A    | 5557G>A  | SOMATIC  | GRCH3.75 | 11      | ATM     | GENE WITH PROTEIN PRODUCT |
| ABC-123 | GF     | 123101  | ACME    | 3      | TRF001300 | SNV    | ambiguous  | ambiguous | SOMATIC  | GRCH3.75 | 11      | ATM     | GENE WITH PROTEIN PRODUCT |
| ABC-123 | GF     | 123101  | ACME    | 4      | TRF001300 | SNV    | 501        | 501     | SOMATIC  | GRCH3.75 | 11      | ATM     | GENE WITH PROTEIN PRODUCT |
| ABC-123 | GF     | 123101  | ACME    | 5      | TRF001300 | SNV    | 51         | 61      | SOMATIC  | GRCH3.75 | 11      | ATM     | GENE WITH PROTEIN PRODUCT |
Terminology Considerations for GFSYM

Where to put the Gene Name?

SDTMIGv3.4, GF Domain Assumption 5

• “For human genetic data, standard nomenclature populated in variable GFSYM must be obtained from the genomic symbol list maintained in the HUGO Gene Nomenclature Committee (HGNC) database (www.genenames.org).”

• Gene Symbols do not belong in GFTEST/CD – Request will be denied.
Terminology Considerations for GFANMETH

• GFANMETH variable is supported by the GFANMET codelist
  • Contains a list of named formulas or gene signatures
  • Codelist is extensible

• The definition for each value will contain a text description of the formula.

• The actual mathematical formula can be placed in the Define-XML file, owing to character constraints in the dataset.

• When submitting a CDISC change request for a new GFANMETH value, a paper citation for the formula as well as the related GFTEST and GFTSTDTL values should be submitted with the request for better understanding by the team.
Putting it together

GF Examples in SDTMIG v3.4

Example 1
Single Nucleotide and Copy Number Variation

Example 2
Single Nucleotide Variation

Example 3
Transcription

Example 4
Microsatellite Instability

Example 5
Microbial Genetics

Shows relationships between GF, MB, and MS
GF Relationship to Pharmacogenomics/Genetics Findings (PF)

Genomics Findings (GF) is continuous improvement of standards.

Study Data Tabulation Model Implementation Guide: Pharmacogenomics/Genetics Version 1.0 (SDTMIG-PGx v1.0)

Refinement of PF lead to development of GF for SDTMIG v3.4
GF Relationship to Pharmacogenomics/Genetics Findings (PF)

Summary

• Domain renamed Genomics Findings (GF) with clarification of scope
• New use cases modeled for GF
• Eighteen variables with overlapping concepts and unclear definitions clarified
• Two new concepts added
• Five established SDTM variables added
• Two outdated concepts retired

Please also find a detailed summary in the Back-up section of this slide deck.
Future Directions
Today

The CDISC Genomics Subteam goal for 2022 is to:

• Support stakeholder implementation of genomics standards through outreach and development/publication of resources and new standards

To achieve this goal, we are working toward deliverables related to:

• Communication of Standards
• Implementation Support
• Standards Development
• Refinements to Genomics Findings (GF)
In progress for 2022

Communication of Standards

- Introduction to the SDTM Genomics Findings (GF) Domain Webinar
- CDISC Europe Interchange (April)
- Additional conference presentations (TBD)
- Training course (TBD)

Implementation Support

- CDISC Website Landing page and FAQs (estimated late 2022)
- Introduction to GF Knowledge Base Article (TBD)
- GF domain examples in Examples Collection (estimated mid to late 2022)
In progress for 2022

Standards Development

• GF Codetable Mapping File (to be published 25 March)
• Controlled Terminology Rules for GF (to be published 24 June)
• CDASH collaboration for genomic data collection (scoping March)

Refinements to Genomics Findings (GF)

• Pending development work (TBD)
Coming Soon

GF Controlled Terminology Development Rules Document

- Enables consistent decision making by Genomics Team; Enables understanding of how GF terminology is built for the user community
- Specific rules around terminology development for GFTEST/CD, GFTSTDTL, GFANMETH
- Describes how GFSYM should be populated with an external terminology
- Will be expanded as the GF terminology matures
- Document currently out for Public Review!
Coming Soon

GF domain examples in Examples Collection

• Initial drafts completed for:
  1. Sequence Rearrangement Fusion
  2. Short Variation Insertions and Deletions
  3. Tumor Mutation Burden
  4. Variable Number of Tandem Repeats

• Drafts in progress:
  5. CYP450
  6. HLA Typing
Tumor Mutation Burden

This example shows findings from an assessment of the number of mutations within a tumor genome with the purpose of determining likely response to a therapeutic agent and/or disease burden. In this example, findings are generated by two vendors using different methodologies and specimen types.

**gf.xpt**

**Row 1:** Shows the number of sequence variants within the region of interest. The vendor, methodology, and specimen type are shown in GFNAM, GFMETHOD, and GFSPEC.

**Row 2:** Shows the normalized number of sequence variants within the region of interest. The panel of genes used in next generation targeted sequencing is shown in SPDVID. The vendor, methodology, and specimen type are shown in GFNAM, GFMETHOD, and GFSPEC.

**Row 3:** Shows the summary of the magnitude of the variant burden within the tumor. The panel of genes used in next generation targeted sequencing is shown in SPDVID. The vendor, methodology, and specimen type are shown in GFNAM, GFMETHOD, and GFSPEC.

<table>
<thead>
<tr>
<th>Row</th>
<th>STUDYID</th>
<th>DOMAIN</th>
<th>USUBJID</th>
<th>SPDVID</th>
<th>GFSEQ</th>
<th>GFREFD</th>
<th>GFTESTCD</th>
<th>GFTEST</th>
<th>GFTSTDTL</th>
<th>GFORRES</th>
<th>GFORRESU</th>
<th>GFSTRESC</th>
<th>GFSTRESN</th>
<th>GFSTRESU</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>ABC-123</td>
<td>GF</td>
<td>ABC123-45-001</td>
<td>1</td>
<td>78975864</td>
<td>TMB</td>
<td>Tumor Mutation Burden</td>
<td>NUMBER OF SEQUENCE VARIANTS</td>
<td>497</td>
<td>497</td>
<td>497</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>ABC-123</td>
<td>GF</td>
<td>ABC123-45-001</td>
<td>2</td>
<td>96757855</td>
<td>TMB</td>
<td>Tumor Mutation Burden</td>
<td>NORMALIZED NUMBER OF SEQUENCE VARIANTS</td>
<td>8.83</td>
<td>/MBP</td>
<td>8.83</td>
<td>8.83</td>
<td>/MBP</td>
<td></td>
</tr>
<tr>
<td>3</td>
<td>ABC-123</td>
<td>GF</td>
<td>ABC123-45-001</td>
<td>3</td>
<td>96757855</td>
<td>TMB</td>
<td>Tumor Mutation Burden</td>
<td>VARIANT SEQUENCE BURDEN INTERPRETATION</td>
<td>INTERMEDIATE</td>
<td>INTERMEDIATE</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
How you can be involved!
How you can be involved!

We invite you to contribute to continuous improvement of genomic standards:

• Become a CDISC Genomics Subteam volunteer
  • [www.cdisc.org/volunteer](http://www.cdisc.org/volunteer)
    • Click link to *Become a Volunteer*

• Contribute FAQs and use case examples for modeling
  • Use cases examples should be real-life, de-identified, and submission related
  • We would like to discuss your use case examples with you
  • Reach out to Christine Connolly, CDISC Senior Project Manager ([cconnolly@cdisc.org](mailto:cconnolly@cdisc.org))

• Review draft standards as they are released
Why volunteer?

Volunteers gain professional experience

Teams bring people together – Networking, etc.

Learn different things about standards and the development process

Volunteering strengthens the standards community

You get a chance to give back and make a difference

Unique opportunity to influence the standard development process
Thank You!
Back-up: Detailed Summary of GF Relationship to PF
Detailed Summary of GF Relationship to PF

<table>
<thead>
<tr>
<th>Domain</th>
<th>Refined domain is named <em>Genomics Findings (GF)</em> to reflect genomic data in scope</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Scope and assumptions clarified and expanded in GF</td>
</tr>
<tr>
<td></td>
<td>New use cases are modeled for GF</td>
</tr>
</tbody>
</table>
## Detailed Summary of GF Relationship to PF

<table>
<thead>
<tr>
<th>Identifier Variables</th>
<th>GF specifies variable SPDEVID and appropriate Medical Device domains may be used to represent the platform used to detect the finding and/or associated assay panels, reagents; as needed</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Variables Non-host Species (PFNPSCES) and Non-host Strain (PFNSTRN) have been replaced by Non-host Organism Identifier (NHOID) in GF</td>
</tr>
<tr>
<td></td>
<td>Established variable Link Group ID (GFLNKGRP) is added in GF</td>
</tr>
</tbody>
</table>
### Detailed Summary of GF Relationship to PF

<table>
<thead>
<tr>
<th>Topic Variable</th>
<th>Values in Short Name of Genomic Measurement (GFTESTCD) and Name of Genomic Measurement (GFTEST) represent the genomic assessment as the topic for the record</th>
</tr>
</thead>
</table>
### Detailed Summary of GF Relationship to PF

<table>
<thead>
<tr>
<th>Qualifier Variables</th>
<th>Values in <strong>Short Name of Genomic Measurement (GFTESTCD)</strong> and <strong>Name of Genomic Measurement (GFTEST)</strong> represent the genomic assessment as the topic for the record</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td><strong>Measurement, Test, or Examination Detail (GFTSTDTL)</strong> represents the reportable from the genomic assessment</td>
</tr>
<tr>
<td></td>
<td><strong>Variables Category for Genomic Finding (GFCAT) and Subcategory for Genomic Finding (GFSCAT)</strong> are sponsor defined.</td>
</tr>
</tbody>
</table>
### Detailed Summary of GF Relationship to PF

#### Qualifier Variables

Variables Result or Finding in Original Units (GFORRES), Result or Finding in Standard Format (GFSTRESC), Numeric Result/Finding in Standard Units (GFSTRESN) follow established Findings Class rules for population; where the value of GFORRES is the original result and GFSTRESC and GFSTRESN are standardized versions of GFORRES where appropriate.

Established variable Reference Result in Standard Format (GFSTREFC) is added in GF.

Established variable Numeric Reference Result in Std Units (GFSTREFN) is added in GF.
### Detailed Summary of GF Relationship to PF

<table>
<thead>
<tr>
<th>Qualifier Variables</th>
<th>Variable Inheritability (GFINHERT) represents whether the variation can be passed to the next generation. Mutation Type (PFMUTYP) is replaced</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>New variable Genome Reference (GFGENREF) is added in GF</td>
</tr>
<tr>
<td></td>
<td>New variable Chromosome (GFCHROM) is added in GF</td>
</tr>
</tbody>
</table>
## Detailed Summary of GF Relationship to PF

<table>
<thead>
<tr>
<th>Qualifier Variables</th>
</tr>
</thead>
<tbody>
<tr>
<td>Variable Genomic Symbol (GFSYM) represents a published symbol for the portion of the genome serving as a locus for the experiment/test. Genetic Region of Interest (PFGENRI) is replaced</td>
</tr>
<tr>
<td>Variable Genomic Symbol Type (GFSYMTYP) represents a description of the type of genomic entity that is represented by the published symbol in GFSYM. Type of Genetic Region of Interest (PFGENTYP) is replaced</td>
</tr>
<tr>
<td>Variable Genetic Location of Interest (PFGENLI) is retired</td>
</tr>
<tr>
<td>Variable Genetic Target (PFGENTRG) is retired</td>
</tr>
</tbody>
</table>
## Detailed Summary of GF Relationship to PF

<table>
<thead>
<tr>
<th>Qualifier Variables</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Variable Sequence Identifier (GFSEQID)</td>
<td>represents a unique identifier for the sequence used as the reference to identify the genetic variation in the result. Reference Sequence (PFREFSEQ) is replaced</td>
</tr>
<tr>
<td>Variable Published Variant Identifier (GFPVRID)</td>
<td>represents a unique identifier for the variation that has been publicly characterized in an external database. Reference SNP Cluster ID Number (PFRSNUM) is replaced</td>
</tr>
<tr>
<td>Variable Copy Identifier (GFCOPYID)</td>
<td>represents an arbitrary identifier used to differentiate between copies of a genetic target of interest present on homologous chromosomes. Allele (Chromosome) Identifier (PFALLELC) is replaced</td>
</tr>
</tbody>
</table>
## Detailed Summary of GF Relationship to PF

| Qualifier Variables | It is specified that variable Method of Test or Examination (GFMETHOD) represents test method by which the examination is performed by the wet lab in order to yield the result reported in the dataset. |
|---------------------|-------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------
|                     | It is specified that variable Analysis Method (GFANMETH) represents the method of secondary processing performed by the dry lab to yield the result reported in the dataset. |
**Detailed Summary of GF Relationship to PF**

<table>
<thead>
<tr>
<th>Timing Variables</th>
<th>Established variable Time Point Reference (GFTPTREF) added in GF</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Established variable Date/Time of Reference Time Point (GFRFTDTC) added in GF</td>
</tr>
</tbody>
</table>
Questions & Answers
Any introduction to key concepts will be helpful for programmers who do not have expertise in genomics.
Audience Questions

We work with NGS data produced by various sequencers. Can we use the new (GF) for Genomic Findings domain for managing NGS data?
Audience Questions

Which of the SDTM Variables require collection in CRFs?
Audience Questions

In addition to VARIANT IMPACT CLASS., how should further types of alteration be mapped in GF (Missense, Non-Frameshift...)?
Do we expect to record cytogenetics findings or any chromosomal abnormalities which may have been determined using methods?
Audience Questions

How is interoperability with both Terra platform and with OMOP standard data pipelines envisioned with SDTM data?
If an organization has been using PF, 1) will it be required to and 2) is it straightforward to shift to using GF in its place?
Audience Questions

We would like a mapping from PF (or other deprecated domains) to GF. Since we have extensively used PF, it does get difficult at time to figure where we would map something originally in PF.
Audience Questions

How would rearrangement for two genes: gene fusions be represented? This is not covered in the examples.
Audience Questions

Need more definition for GENLOC. For results from cytogenetics, we often see location ranges. How would you represent that in Location? Some more examples from cytogenetics and NGS would be helpful.
We would like more definition for GENLOC. Can you provide examples for cyogenetics where one or more chromosomes and related locations or location ranges?
Re: example 1 (from Glenn showing the wiki): how would more granular read depths be represented? i.e. read depths need to be linked to specific nucleotides within one gene?
Still Re: example 1, row 8: How should the reference gene copy number be represented based on which the copy number ratio is built?
Audience Questions

How would a rearrangement between two genes detected by NGS be expected to be reported (e.g. EML4-ALK)?
Audience Questions

GFINHERT: Should this be Y or N only?
Audience Questions

For GFGENLOC, what are the possible regions available? Exon? Base pair to base pair? Etc
Audience Questions

For GFGENLOC, what are the possible regions available? Exon? Base pair to base pair? Etc
One example that's lacking in SDTMIG 3.4 is for a translocation/gene fusion. Could you please explain how genetic findings involving two different loci should be represented?
Re: Example 3 (transcription): How should we represent which housekeeping gene is used for gene expression?
Audience Questions

GFTEST and GFTESTCD is not very unique in this domain. Why not create unique Test codes / Test names?
Maybe a bit off-topic, but just out of interest given you have mentioned gene names will never be part of GFTESTCD anymore: is CDISC planning to take the same approach for MITESTCD as well? MITESTCD terminology is not easily manageable with the current approach because protein names are included there.
Audience Questions

New examples and draft will be very useful!
Upcoming Learning Opportunities
April - May 2022
Europe Interchange Trainings

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The OpenStudyBuilder is an open-source project for clinical study specification. This tool is a new approach for working with studies that once fully implemented will drive end-to-end consistency and more efficient processes - all the way from protocol development and CRF design - to creation of datasets, analysis, reporting, submission to health authorities and public disclosure of study information.
# Upcoming Webinars

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Ideas or suggestions for webinar topics? Any topics you would love to see us cover?

Let us know via our topic suggestion form:
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