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

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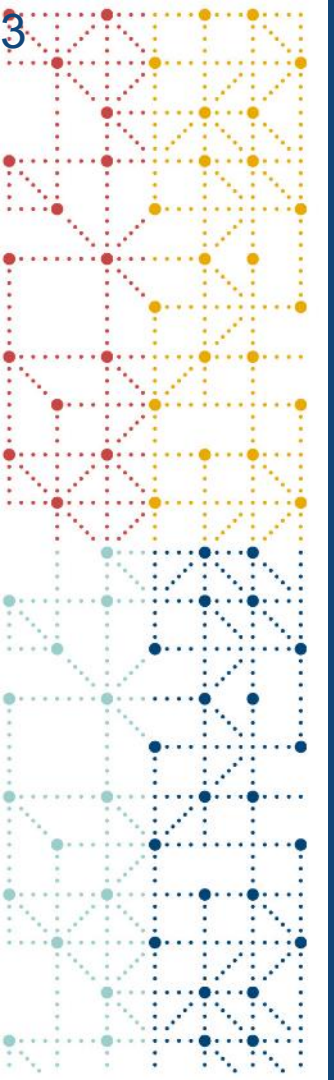
THU 24 MAR 2022
11:00AM-12:30PM ET

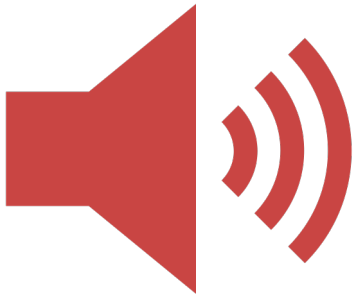
Today's Agenda

1. Housekeeping
2. Speaker Introductions
3. Feature Presentation
4. Upcoming Learning Opportunities & Events



Housekeeping

Housekeeping

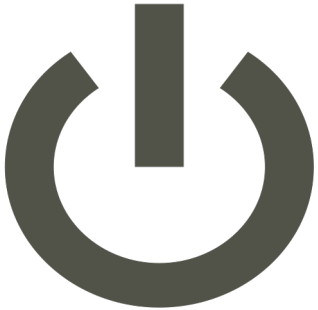


You will remain on **mute**

Housekeeping

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

Housekeeping

A recording of this webinar and the slides will be available in the **Members Only** section of CDISC website

Today's Presenters

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
Enterprise Vocabulary Services
National Cancer Institute

Jon Neville
Senior Standards Developer
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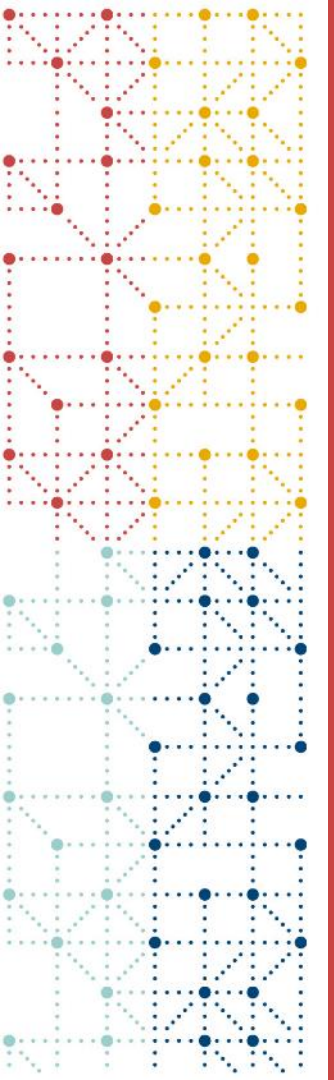
Genomics

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

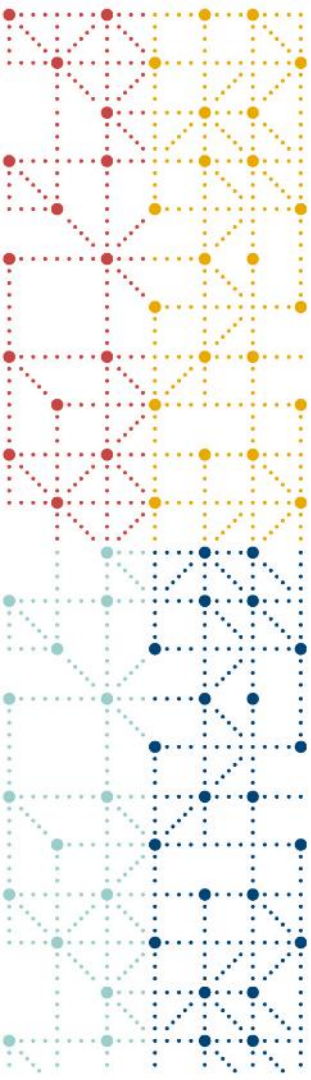
Genomic data collected as part of clinical research supports both development of quality patient care and improvements in patient outcomes.

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!



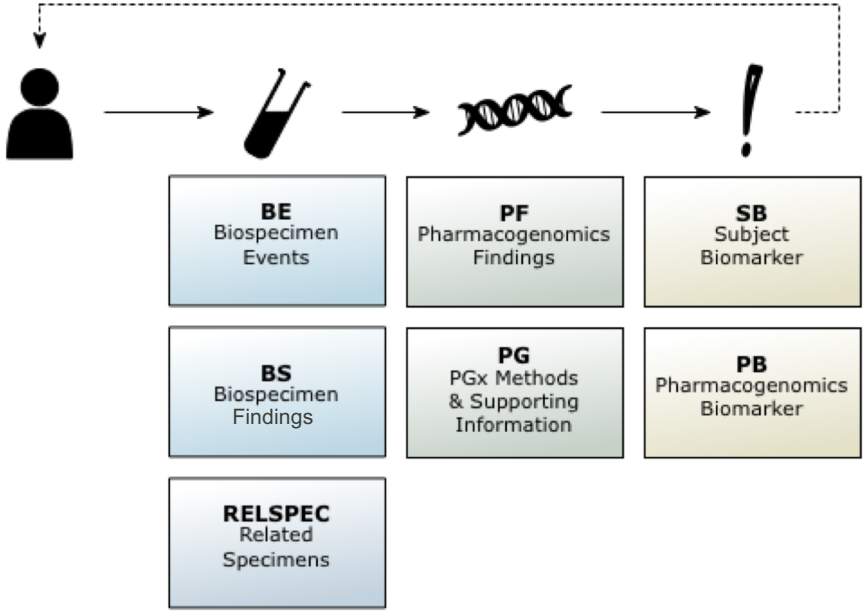
History

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

SDTMIG-PGx v1.0

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

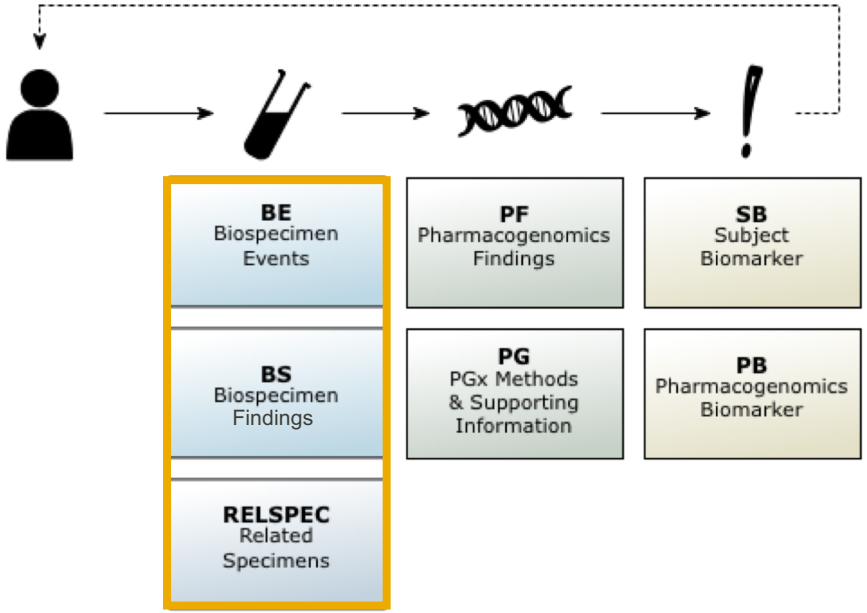
A single domain, Genomics Findings (GF), published in the SDTMIG v3.4 in 2021

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

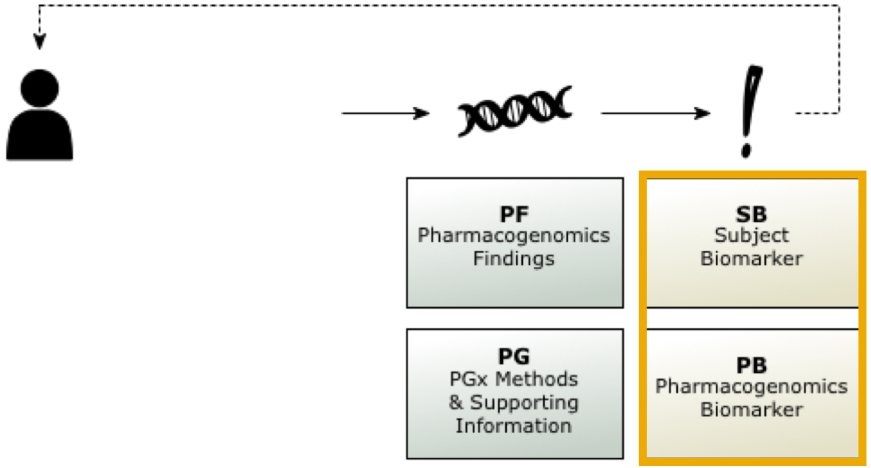
A *single domain*, Genomics Findings (GF), published in the SDTMIG v3.4



- 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

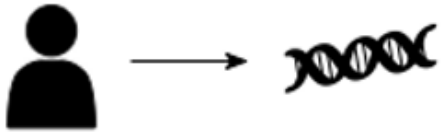
A *single domain*, Genomics Findings (GF), published in the SDTMIG v3.4



- 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



PF
Pharmacogenomics Findings

PG
PGx Methods & Supporting Information

- 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

PF
Pharmacogenomics Findings

- 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

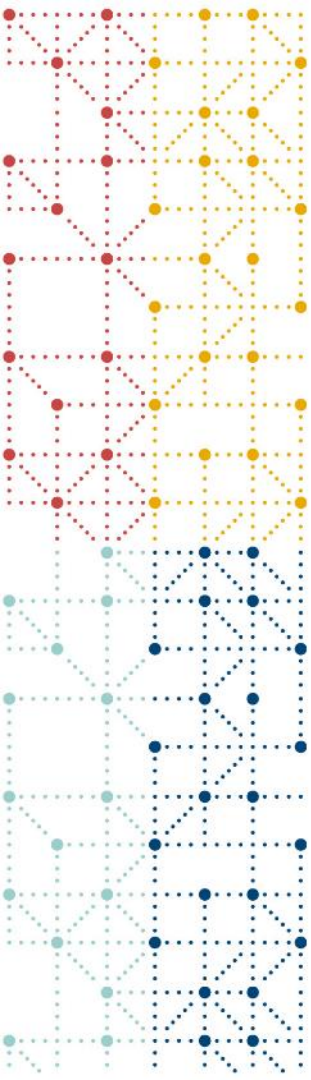
Rationale

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.

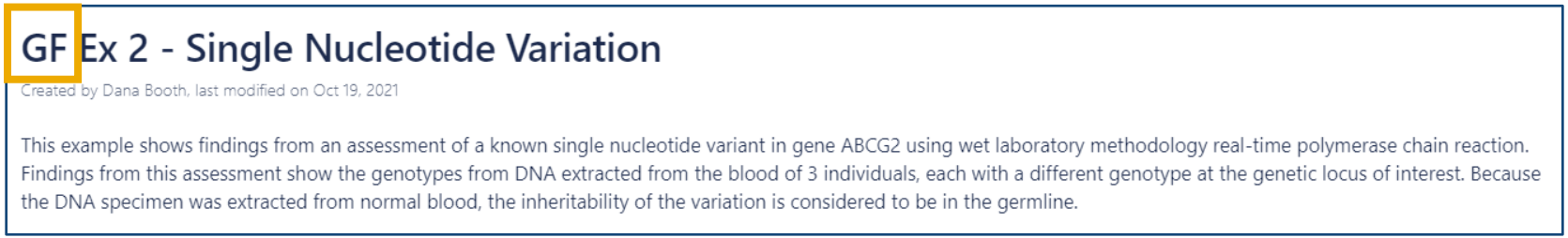
gf.xpt

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.

gf.xpt

Row	STUDYID	DOMAIN	USUBJID	GFSEQ	GFREFID	GFTESTCD	GFTEST	GFTSTDTL	GFORRES	GFORREF	GFSTRESC	GFSTREFC	GFINHERT	GFGENREF	GFCHROM	GFSYM
1	C12345	GF	C12345-001	1	NA18537	SNV	Single Nucleotide Variation	GENOTYPE	T/T	G/G	T/T	G/G	GERMLINE VARIATION	GRCh38.p13	4	ABCG2

GF Domain Scope



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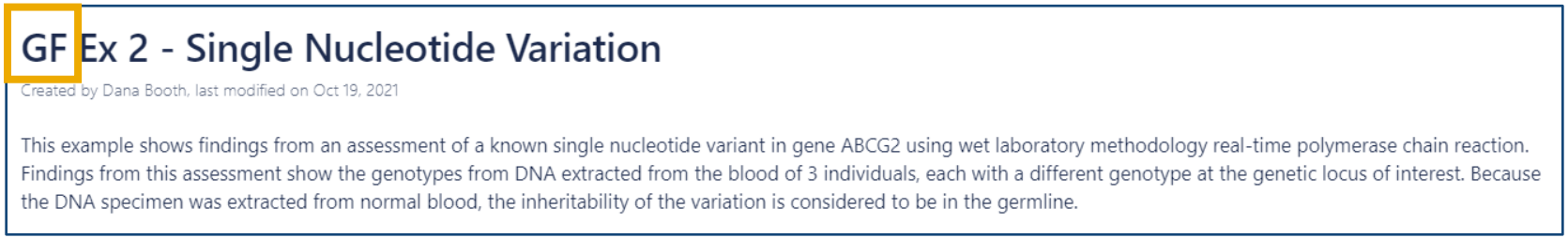
Representation of findings related to the structure, function, evolution, mapping, and editing of subject and non-host organism genomic material of interest; i.e.,:

- 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



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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 Record Structure

GF Ex 2 - Single Nucleotide Variation

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1	C12345	GF	C12345-001	1	NA18537	SNV	Single Nucleotide Variation	GENOTYPE	T/T	G/G	T/T	G/G	GERMLINE VARIATION	GRCh38.p13	4	ABCG2

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

GF Variables

GF Ex 2 - Single Nucleotide Variation

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1	C12345	GF	C12345-001	1	NA18537	SNV	Single Nucleotide Variation	GENOTYPE	T/T	G/G	T/T	G/G	GERMLINE VARIATION	GRCh38.p13	4	ABCG2

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

GF Identifier Variables

GF Ex 2 - Single Nucleotide Variation

STUDYID	DOMAIN	USUBJID	GFSEQ	GFREFID	GFTESTCD	GFTEST	GFTSTDTL	GFORRES
C12345	GF	C12345-001	1	NA18537	SNV	Single Nucleotide Variation	GENOTYPE	T/T

Variable Name	Variable Label	Type	Controlled Terms, Codelist or Format[1]	Role	CDISC Notes	Core
STUDYID ★	Study Identifier	Char		Identifier	Unique identifier for a study.	Req
DOMAIN ★	Domain Abbreviation	Char	GF	Identifier	Two-character abbreviation for the domain.	Req
USUBJID ★	Unique Subject Identifier	Char		Identifier	Identifier used to uniquely identify a subject across all studies for all applications or submissions involving the product.	Req
SPDEVID ★	Sponsor Device Identifier	Char		Identifier	Sponsor-defined identifier for a device.	Perm
NHOID ★	Non-Host Organism Identifier	Char		Identifier	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., "A/California/7/2009 (H1N1)"). It is not to be used as a qualifier of the result in the record on which it appears.	Perm
GFSEQ ★	Sequence Number	Num		Identifier	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.	Req
GFGRPID ★	Group ID	Char		Identifier	Used to link together a block of related records within a subject in a domain.	Perm
GFREFID ★	Reference ID	Char		Identifier	A unique identifier for the assayed genetic specimen.	Exp
GFSPID ★	Sponsor-Defined Identifier	Char		Identifier	Sponsor-defined identifier.	Perm
GFLNKID ★	Link ID	Char		Identifier	Identifier used to link related records across domains. This may be a one-to-one or a one-to-many relationship.	Perm
GFLNKGRP ★	Link Group ID	Char		Identifier	Identifier used to link related records across domains. This will usually be a many-to-one relationship.	Perm

Platform used to detect the finding may be represented here

GF Topic Variable

GF Ex 2 - Single Nucleotide Variation

STUDYID	DOMAIN	USUBJID	GFSEQ	GFREFID	GFTESTCD	GFTEST	GFTSTDTL	GFORRES
C12345	GF	C12345-001	1	NA18537	SNV	Single Nucleotide Variation	GENOTYPE	T/T

Variable Name	Variable Label	Type	Controlled Terms, Codelist or Format[1]	Role	CDISC Notes	Core
GFTESTCD ★	Short Name of Genomic Measurement	Char	(GFTESTCD)	Topic	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., "1TEST" is not valid). GFTESTCD cannot contain characters other than letters, numbers, or underscores.	Req

GF Qualifier Variables

GF Ex 2 - Single Nucleotide Variation

STUDYID	DOMAIN	USUBJID	GFSEQ	GFREFID	GFTESTCD	GFTEST	GFTSTDTL	GFORRES
C12345	GF	C12345-001	1	NA18537	SNV	Single Nucleotide Variation	GENOTYPE	T/T

Variable Name	Variable Label	Type	Controlled Terms, Codelist or Format[1]	Role	CDISC Notes	Core
GFTEST ★	Name of Genomic Measurement	Char	(GFTEST)	Synonym Qualifier	Long name for GFTESTCD. The value in GFTEST cannot be longer than 40 characters.	Req
GFTSTDTL ★	Measurement, Test, or Examination Detail	Char	(GFTSDTL)	Variable Qualifier	Description of a reportable qualifying the assessment in GFTESTCD and GFTEST.	Perm
GFCAT ★	Category for Genomic Finding	Char		Grouping Qualifier	Used to define a category of topic-variable values.	Perm
GFSCAT ★	Subcategory for Genomic Finding	Char		Grouping Qualifier	Used to define a further categorization of GFCAT values.	Perm

GF Qualifier Variables

GF Ex 2 - Single Nucleotide Variation

GFTESTCD	GFTEST	GFTSTDTL	GFORRES	GFORREF	GFSTRESC	GFSTREFC	GFINHERT	GFGENREF	GFCHROM	GFSYM	GFSYMTYP	GFGENLOC	GFSEQID	GFPVRID	GFNAM	GFSPEC	GFMETHOD
SNV	Single Nucleotide Variation	GENOTYPE	T/T	G/G	T/T	G/G	GERMLINE VARIATION	GRCh38.p13	4	ABCG2	GENE WITH PROTEIN PRODUCT	4:88131171	ENSG00000118777	rs2231142	ACME LABS	DNA	REAL_TIME POLYMERASE CHAIN REACTION

Variable Name	Variable Label	Type	Controlled Terms, Codelist or Format[1]	Role	CDISC Notes	Core
GFORRES ★	Result or Finding in Original Units	Char		Result Qualifier	Result of the measurement or finding as originally received or collected.	Exp
GFORRESU ★	Original Units	Char	(UNIT)	Variable Qualifier	Unit for GFORRES.	Perm
GFORREF ★	Reference Result in Original Units	Char		Variable Qualifier	Reference value for the result or finding as originally received or collected. GFORREF uses the same units as GFORRES, if applicable.	Perm
GFSTRESC ★	Result or Finding in Standard Format	Char		Result Qualifier	Contains the result value for all findings, copied or derived from GFORRES, in a standard format or in standard units. GFSTRESC should store all results or findings in character format; if results are numeric, they should also be stored in numeric format in GFSTRESN.	Exp
GFSTRESN ★	Numeric Result/Finding in Standard Units	Num		Result Qualifier	Used for continuous or numeric results or findings in standard format; copied in numeric format from GFSTRESC. GFSTRESN should store all numeric test results or findings.	Perm
GFSTRESU ★	Standard Units	Char	(UNIT)	Variable Qualifier	Standardized units used for GFSTRESC, GFSTRESN, GFSTREFC, and GFSTREFN.	Perm
GFSTREFC ★	Reference Result in Standard Format	Char		Variable Qualifier	Reference value for the result or finding copied or derived from GFORREF in a standard format.	Perm
GFSTREFN ★	Numeric Reference Result in Std Units	Num		Variable Qualifier	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.	Perm
GFRESCAT ★	Result Category	Char		Variable Qualifier	Used to categorize the result of a finding.	Perm

GF Qualifier Variables

GF Ex 2 - Single Nucleotide Variation

GFTESTCD	GFTEST	GFTSTDTL	GFORRES	GFORREF	GFSTRESC	GFSTREFC	GFINHERT	GFGENREF	GFCHROM	GFSYM	GFSYMTYP	GFGENLOC	GFSEQID	GFPVRID	GFNAM	GFSPEC	GFMETHOD
SNV	Single Nucleotide Variation	GENOTYPE	T/T	G/G	T/T	G/G	GERMLINE VARIATION	GRCh38.p13	4	ABCG2	GENE WITH PROTEIN PRODUCT	4:88131171	ENSG00000118777	rs2231142	ACME LABS	DNA	REAL_TIME POLYMERASE CHAIN REACTION

New variables for genomics in SDTM v2.0

Variable Name	Variable Label	Type	Controlled Terms, Codelist or Format[1]	Role	CDISC Notes	Core
GFINHERT ★	Inheritability	Char	(INHERTGF)	Variable Qualifier	Identifies whether the variation can be passed to the next generation.	Perm
GFGENREF ★	Genome Reference	Char		Variable Qualifier	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 "GRCh38.p13".	Perm
GFCHROM ★	Chromosome Identifier	Char		Variable Qualifier	The designation (name or number) of the chromosome or contig on which the variant or other feature appears (e.g., "17"; "X").	Perm
GFSYM ★	Genomic Symbol	Char	*	Variable Qualifier	A published symbol for the portion of the genome serving as a locus for the experiment/test.	Perm
GFSYMTYP ★	Genomic Symbol Type	Char	(SYMTYPGF)	Variable Qualifier	A description of the type of genomic entity that is represented by the published symbol in GFSYM.	Perm
GFGENLOC ★	Genetic Location	Char		Variable Qualifier	Specifies the location within a sequence for the observed value in GFORRES.	Perm
GFGENSR ★	Genetic Sub-Region	Char		Variable Qualifier	The portion of the locus in which the variation was found. Examples: "Exon 15", "Kinase domain".	Perm
GFSEQID ★	Sequence Identifier	Char		Variable Qualifier	A unique identifier for the sequence used as the reference to identify the genetic variation in the result. Examples: "NM_001234", "ENSG00000182533", "ENST00000343849.2".	Perm
GFPVRID ★	Published Variant Identifier	Char		Variable Qualifier	A unique identifier for the variation that has been publicly characterized in an external database. Examples: "rs2231142", "COSM41596".	Perm
GFCOPYID ★	Copy Identifier	Char		Variable Qualifier	An arbitrary identifier used to differentiate between copies of a genetic target of interest present on homologous chromosomes.	Perm

GF Qualifier Variables

GF Ex 2 - Single Nucleotide Variation

GFTESTCD	GFTEST	GFTSTDTL	GFORRES	GFORREF	GFSTRESC	GFSTREFC	GFINHERT	GFGENREF	GFCHROM	GFSYM	GFSYMTYP	GFGENLOC	GFSEQID	GFPVRID	GFNAM	GFSPEC	GFMETHOD
SNV	Single Nucleotide Variation	GENOTYPE	T/T	G/G	T/T	G/G	GERMLINE VARIATION	GRCh38.p13	4	ABCG2	GENE WITH PROTEIN PRODUCT	4:88131171	ENSG00000118777	rs2231142	ACME LABS	DNA	REAL_TIME POLYMERASE CHAIN REACTION

Variable Name	Variable Label	Type	Controlled Terms, Codelist or Format[1]	Role	CDISC Notes	Core
GFSTAT ★	Completion Status	Char	(ND)	Record Qualifier	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 "NOT DONE".	Perm
GFREASND ★	Reason Test Not Done	Char		Record Qualifier	Reason not done. Used in conjunction with GFSTAT when value is "NOT DONE".	Perm
GFXFN ★	External File Path	Char		Record Qualifier	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.	Perm
GFNAM ★	Laboratory/Vendor Name	Char		Record Qualifier	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.	Perm
GFSPEC ★	Specimen Material Type	Char	(GENSMP)	Record Qualifier	Identifies the type of genetic material used for the measurement.	Perm

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GF Qualifier Variables

GF Ex 2 - Single Nucleotide Variation

GFTESTCD	GFTEST	GFTSTDTL	GFORRES	GFORREF	GFSTRESC	GFSTREFC	GFINHERT	GFGENREF	GFCHROM	GFSYM	GFSYMTYP	GFGENLOC	GFSEQID	GFPVRID	GFNAM	GFSPEC	GFMETHOD
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Variable Name	Variable Label	Type	Controlled Terms, Codelist or Format[1]	Role	CDISC Notes	Core
GFMETHOD ★	Method of Test or Examination	Char	(METHOD)	Record Qualifier	The test method by which the examination is performed by the wet lab in order to yield the result reported in the dataset.	Exp
GFRUNID ★	Run ID	Char		Record Qualifier	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.	Perm
GFANMETH ★	Analysis Method	Char	(GFANMET)	Record Qualifier	The method of secondary processing performed by the dry lab to yield the result reported in the dataset.	Perm
GFBLFL ★	Baseline Flag	Char	(NY)	Record Qualifier	Indicator used to identify a baseline value. Should be "Y" or null.	Perm
GFDRVFL ★	Derived Flag	Char	(NY)	Record Qualifier	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.	Perm
GFLLOQ ★	Lower Limit of Quantitation	Num		Variable Qualifier	Indicates the lower limit of quantitation for an assay. Units will be those used for GFSTRESU.	Perm
GFREPNUM ★	Repetition Number	Num		Record Qualifier	The instance number of a test that is repeated within a given timeframe for the same test performed by the wet lab.	Perm

GF Timing Variables

GF Ex 2 - Single Nucleotide Variation

VISITNUM	VISIT	VISITDY	GFDTC	GFDY
1	SCREENING	-1	2020-06-25	-3

Variable Name	Variable Label	Type	Controlled Terms, Codelist or Format[1]	Role	CDISC Notes	Core
VISITNUM ★	Visit Number	Num		Timing	Clinical encounter number. Numeric version of VISIT, used for sorting.	Exp
VISIT ★	Visit Name	Char		Timing	Protocol-defined description of clinical encounter.	Perm
VISITDY ★	Planned Study Day of Visit	Num		Timing	Planned study day of VISIT. Should be an integer.	Perm
GFDTC ★	Date/Time of Specimen Collection	Char	ISO 8601 datetime or interval	Timing	Date and time of specimen collection.	Exp
GFDY ★	Study Day of Specimen Collection	Num		Timing	Actual study day of visit/collection/exam expressed in integer days relative to the sponsor-defined RFSTDTC in Demographics.	Perm
GFTPT ★	Planned Time Point Name	Char		Timing	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.	Perm
GFTPTNUM ★	Planned Time Point Number	Num		Timing	Numerical version of GFTPT used in sorting.	Perm
GFELTM ★	Planned Elapsed Time from Time Point Ref	Char	ISO 8601 duration	Timing	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.	Perm
GFTPTREF ★	Time Point Reference	Char		Timing	Name of the fixed reference point referred to by GFELTM, GFTPTNUM, and GFTPT. Examples: "PREVIOUS DOSE", "PREVIOUS MEAL".	Perm
GFRFTDTC ★	Date/Time of Reference Time Point	Char	ISO 8601 datetime or interval	Timing	Date/time for a fixed reference time point defined by GFTPTREF.	Perm

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Terminology Considerations

GF variables with Controlled Terminology

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

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 GFTSTDTL

This example shows findings from an assessment of a known single nucleotide variant in gene ABCG2 using wet laboratory methodology real-time polyerase 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 inheritabilty of the variation is considered to be in the germline.

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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.

GF Ex 2 - Single Nucleotide Variation

gf.xpt

Row	STUDYID	DOMAIN	USUBJID	GFSEQ	GFREFID	GFTESTCD	GFTEST	GFTSTDTL	GFORRES	GFORREF	GFSTRESC	GFSTREFC	GFINHERT	GFGENREF	GFCHROM	GFSYM	GFSYMTYP	GFGENLOC	GFSEQID	GFPVRID	GFNAM	GFSPEC	GFMETHOD	VISITNUM	VISIT	VISITDY	GFDTC	GFDY
1	C12345	GF	C12345-001	1	NA1853	SNV	Single Nucleotide Variation	GENOTYPE	T/T	G/G	T/T	G/G	GERMLINE VARIATION	GRCh38.p13	4	ABCG2	GENE WITH PROTEIN PRODUCT	4:88131171	ENSG00000118777	rs2231142	ACME LABS	DNA	REAL_TIME POLYMERASE CHAIN REACTION	1	SCREENING	-1	2020-06-25	-3
2	C12345	GF	C12345-002	2	NA0700	SNV	Single Nucleotide Variation	GENOTYPE	G/T	G/G	G/T	G/G	GERMLINE VARIATION	GRCh38.p13	4	ABCG2	GENE WITH PROTEIN PRODUCT	4:88131171	ENSG00000118777	rs2231142	ACME LABS	DNA	REAL_TIME POLYMERASE CHAIN REACTION	1	SCREENING	-1	2020-06-25	-3
3	C12345	GF	C12345-003	3	NA0013	SNV	Single Nucleotide Variation	GENOTYPE	G/G	G/G	G/G	G/G	GERMLINE VARIATION	GRCh38.p13	4	ABCG2	GENE WITH PROTEIN PRODUCT	4:88131171	ENSG00000118777	rs2231142	ACME LABS	DNA	REAL_TIME POLYMERASE CHAIN REACTION	1	SCREENING	-1	2020-06-25	-3

gf.xpt

Row	STUDYID	DOMAIN	USUBJID	SPDEVID	GFSEQ	GFGRPID	GFREFID	GFTESTCD	GFTEST	GFTSTDTL	GFORRES	GFORRESU	GFSTRESC	GFSTRESN	GFSTRESU	GFINHERT	GFGENREF	GFCHROM	GFSYM	GFSYMTYP	GFGENLOC
1	ABC-123	GF	123101	ACME GenePanel 500	1	1	TRF001338	SNV	Single Nucleotide Variation	PREDICTED AMINO ACID CHANGE	D1853N		D1853N			SOMATIC VARIATION	GRCh37.75	11	ATM	GENE WITH PROTEIN PRODUCT	108175462
2	ABC-123	GF	123101	ACME GenePanel 500	2	1	TRF001338	SNV	Single Nucleotide Variation	PREDICTED CODING SEQUENCE CHANGE	5557G>A		5557G>A			SOMATIC VARIATION	GRCh37.75	11	ATM	GENE WITH PROTEIN PRODUCT	108175462
3	ABC-123	GF	123101	ACME GenePanel 500	3	1	TRF001338	SNV	Single Nucleotide Variation	VARIANT IMPACT CLASSIFICATION	ambiguous		ambiguous			SOMATIC VARIATION	GRCh37.75	11	ATM	GENE WITH PROTEIN PRODUCT	108175462
4	ABC-123	GF	123101	ACME GenePanel 500	4	1	TRF001338	SNV	Single Nucleotide Variation	READ DEPTH	501		501	501		SOMATIC VARIATION	GRCh37.75	11	ATM	GENE WITH PROTEIN PRODUCT	108175462
5	ABC-123	GF	123101	ACME GenePanel 500	5	1	TRF001338	SNV	Single Nucleotide Variation	VARIANT READ DEPTH/READ DEPTH	51	%	51	51	%	SOMATIC VARIATION	GRCh37.75	11	ATM	GENE WITH PROTEIN PRODUCT	108175462

Terminology Considerations for GFSYM

Where to put the Gene Name?

SDTMIGv3.4, GF Domain Assumption 5

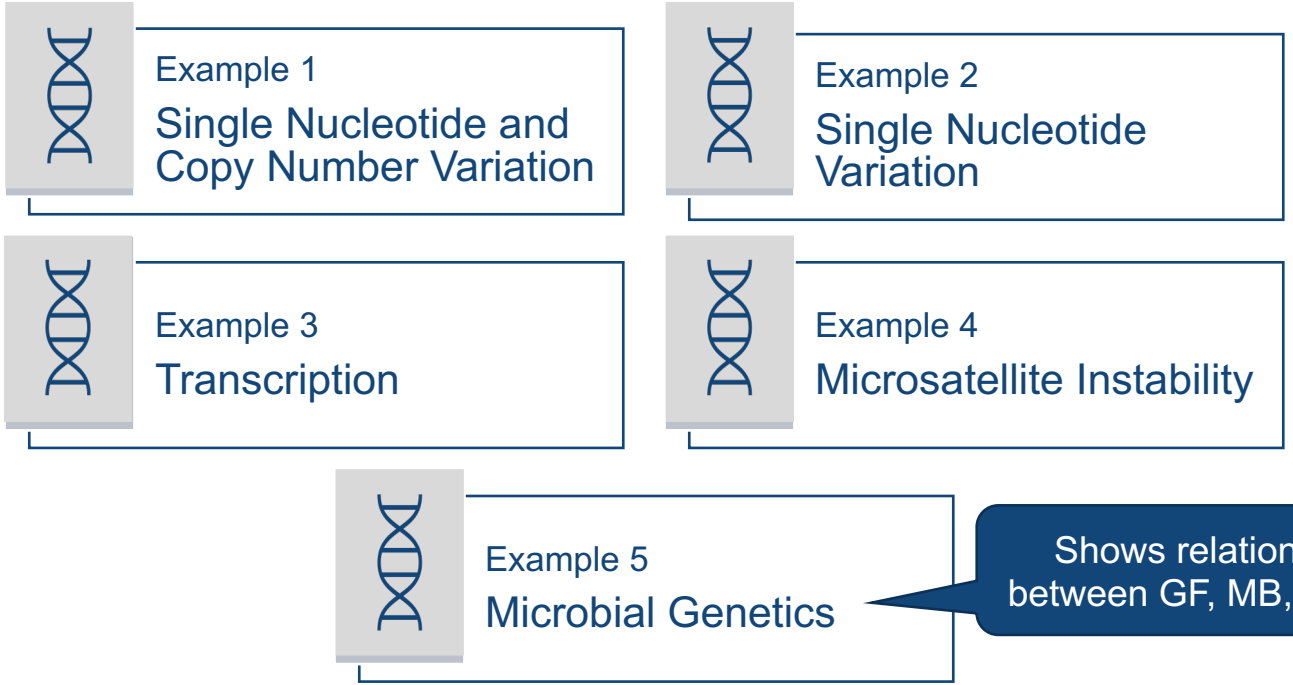
- "For human genetic data, standard nomenclature populated in variable GFSYM must be obtained from the genomic symbol list maintained in the HUGO Gene Nomenclature Committee (HGNC) database (www.genenames.org)."
- Gene Symbols do not belong in GFTEST/CD – *Request will be denied.*

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.

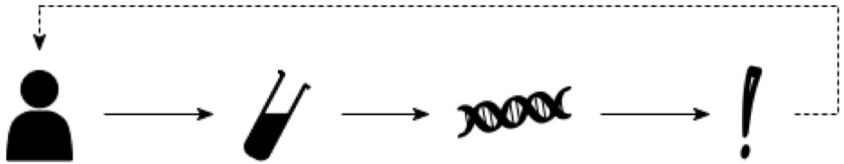
Putting it together

GF Examples in SDTMIG v3.4



GF Relationship to Pharmacogenomics/Genetics Findings (PF)

Genomics Findings (GF) is continuous improvement of standards.



BE Biospecimen Events	**PF** Pharmacogenomics Findings	**SB** Subject Biomarker
BS Biospecimen Findings	**PG** PGx Methods & Supporting Information	**PB** Pharmacogenomics Biomarker
RELSPEC Related Specimens		

Refinement of PF lead to development of GF for SDTMIG v3.4

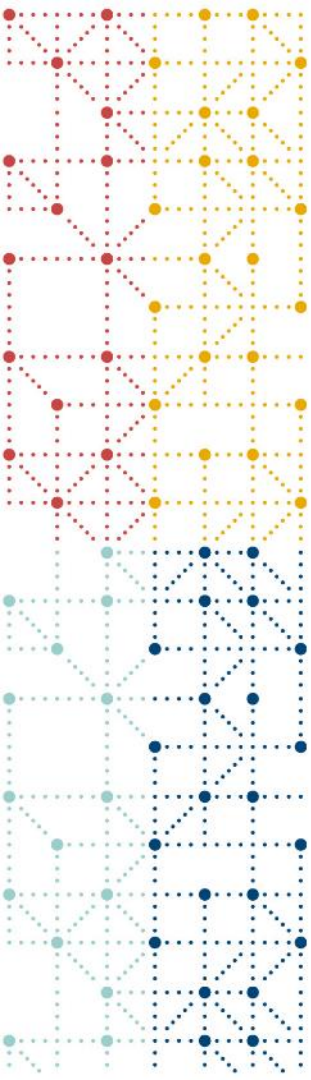
Study Data Tabulation Model Implementation Guide: Pharmacogenomics/Genetics Version 1.0 (SDTMIG-PGx v1.0)

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)

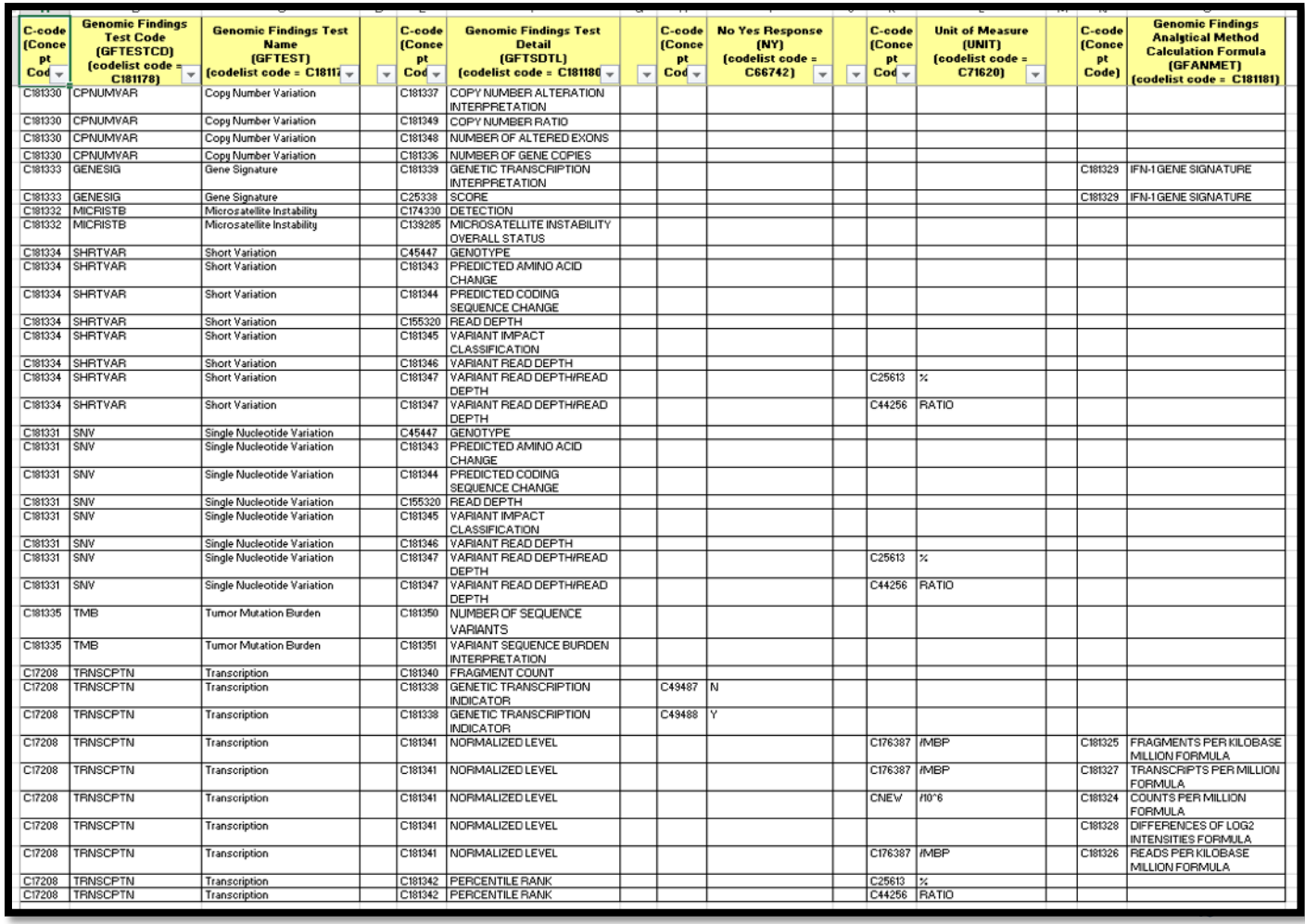
Refinements to Genomics Findings (GF)

- Pending development work (TBD)

Coming Soon

GF Codetable Mapping File

Publication
25 March



C-code (Concept Code)	Genomic Findings Test Code (GFTESTCD) (codelist code = C181178)	Genomic Findings Test Name (GFTEST) (codelist code = C181177)	C-code (Concept Code)	Genomic Findings Test Detail (GFTSDTL) (codelist code = C181180)	C-code (Concept Code)	No Yes Response (NY) (codelist code = C66742)	C-code (Concept Code)	Unit of Measure (UNIT) (codelist code = C71620)	C-code (Concept Code)	Genomic Findings Analytical Method Calculation Formula (GFANMET) (codelist code = C181181)
C181330	CPNUMVAR	Copy Number Variation	C181337	COPY NUMBER ALTERATION INTERPRETATION						
C181330	CPNUMVAR	Copy Number Variation	C181349	COPY NUMBER RATIO						
C181330	CPNUMVAR	Copy Number Variation	C181348	NUMBER OF ALTERED EXONS						
C181330	CPNUMVAR	Copy Number Variation	C181336	NUMBER OF GENE COPIES						
C181333	GENESIG	Gene Signature	C181339	GENETIC TRANSCRIPTION INTERPRETATION					C181329	IFN-1 GENE SIGNATURE
C181333	GENESIG	Gene Signature	C25338	SCORE					C181329	IFN-1 GENE SIGNATURE
C181332	MICRISTB	Microsatellite Instability	C174330	DETECTION						
C181332	MICRISTB	Microsatellite Instability	C139285	MICROSATELLITE INSTABILITY OVERALL STATUS						
C181334	SHRTVAR	Short Variation	C45447	GENOTYPE						
C181334	SHRTVAR	Short Variation	C181343	PREDICTED AMINO ACID CHANGE						
C181334	SHRTVAR	Short Variation	C181344	PREDICTED CODING SEQUENCE CHANGE						
C181334	SHRTVAR	Short Variation	C155320	READ DEPTH						
C181334	SHRTVAR	Short Variation	C181345	VARIANT IMPACT CLASSIFICATION						
C181334	SHRTVAR	Short Variation	C181346	VARIANT READ DEPTH						
C181334	SHRTVAR	Short Variation	C181347	VARIANT READ DEPTH/READ DEPTH			C25613	%		
C181334	SHRTVAR	Short Variation	C181347	VARIANT READ DEPTH/READ DEPTH			C44256	RATIO		
C181331	SNV	Single Nucleotide Variation	C45447	GENOTYPE						
C181331	SNV	Single Nucleotide Variation	C181343	PREDICTED AMINO ACID CHANGE						
C181331	SNV	Single Nucleotide Variation	C181344	PREDICTED CODING SEQUENCE CHANGE						
C181331	SNV	Single Nucleotide Variation	C155320	READ DEPTH						
C181331	SNV	Single Nucleotide Variation	C181345	VARIANT IMPACT CLASSIFICATION						
C181331	SNV	Single Nucleotide Variation	C181346	VARIANT READ DEPTH						
C181331	SNV	Single Nucleotide Variation	C181347	VARIANT READ DEPTH/READ DEPTH			C25613	%		
C181331	SNV	Single Nucleotide Variation	C181347	VARIANT READ DEPTH/READ DEPTH			C44256	RATIO		
C181335	TMB	Tumor Mutation Burden	C181350	NUMBER OF SEQUENCE VARIANTS						
C181335	TMB	Tumor Mutation Burden	C181351	VARIANT SEQUENCE BURDEN INTERPRETATION						
C17208	TRNSCPTN	Transcription	C181340	FRAGMENT COUNT						
C17208	TRNSCPTN	Transcription	C181338	GENETIC TRANSCRIPTION INDICATOR	C49487	N				
C17208	TRNSCPTN	Transcription	C181338	GENETIC TRANSCRIPTION INDICATOR	C49488	Y				
C17208	TRNSCPTN	Transcription	C181341	NORMALIZED LEVEL			C176387	/MBP	C181325	FRAGMENTS PER KILOBASE MILLION FORMULA
C17208	TRNSCPTN	Transcription	C181341	NORMALIZED LEVEL			C176387	/MBP	C181327	TRANSCRIPTS PER MILLION FORMULA
C17208	TRNSCPTN	Transcription	C181341	NORMALIZED LEVEL			CNEW	/10^6	C181324	COUNTS PER MILLION FORMULA
C17208	TRNSCPTN	Transcription	C181341	NORMALIZED LEVEL					C181328	DIFFERENCES OF LOG2 INTENSITIES FORMULA
C17208	TRNSCPTN	Transcription	C181341	NORMALIZED LEVEL			C176387	/MBP	C181326	READS PER KILOBASE MILLION FORMULA
C17208	TRNSCPTN	Transcription	C181342	PERCENTILE RANK			C25613	%		
C17208	TRNSCPTN	Transcription	C181342	PERCENTILE RANK			C44256	RATIO		

Coming Soon

GF Controlled Terminology Development Rules Document


CDISC CONTROLLED TERMINOLOGY RULES:
Genomics Findings Domain (GF)

18 Mar 2022

- 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

Coming Soon

GF domain examples in Examples Collection; draft example

Tumor Mutation Burden

Created by Christine Connolly, last modified on Mar 17, 2022

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.

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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 a 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.

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Row	STUDYID	DOMAIN	USUBJID	SPDEVID	GFSEQ	GFREFID	GFTESTCD	GFTEST	GFTSTDTL	GFORRES	GFORRESU	GFSTRESC	GFSTRESN	GFSTRESU
1	ABC-123	GF	ABC123-45-001		1	78975864	TMB	Tumor Mutation Burden	NUMBER OF SEQUENCE VARIANTS	497		497	497	
2	ABC-123	GF	ABC123-45-001	ACME 500 GENE PANEL	2	96757855	TMB	Tumor Mutation Burden	NORMALIZED NUMBER OF SEQUENCE VARIANTS	8.83	/MBP	8.83	8.83	/MBP
3	ABC-123	GF	ABC123-45-001	ACME 500 GENE PANEL	3	96757855	TMB	Tumor Mutation Burden	VARIANT SEQUENCE BURDEN INTERPRETATION	INTERMEDIATE		INTERMEDIATE		

How you can be involved!

How you can be involved!

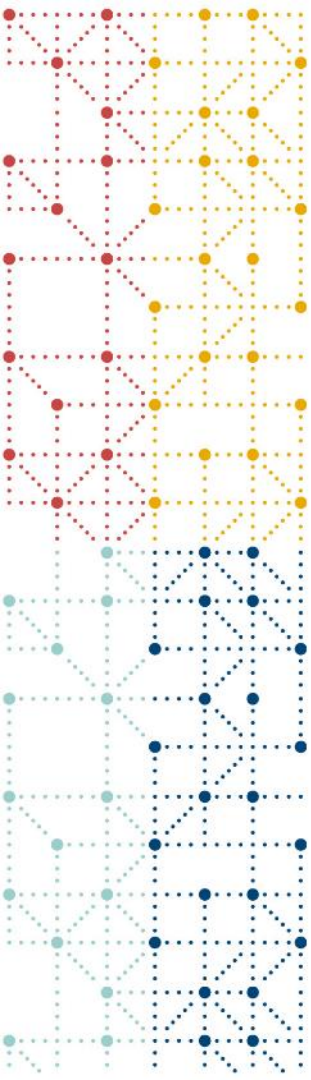
We invite you to contribute to continuous improvement of genomic standards:

- Become a CDISC Genomics Subteam volunteer
  - www.cdisc.org/volunteer
    - 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)
- 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

Domain	Refined domain is named *Genomics Findings (GF)* to reflect genomic data in scope
	Scope and assumptions clarified and expanded in GF
	New use cases are modeled for GF

Detailed Summary of GF Relationship to PF

Identifier Variables	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
	Variables Non-host Species (PFNPSCES) and Non-host Strain (PFNSTRN) have been replaced by Non-host Organism Identifier (NHOID) in GF
	Established variable Link Group ID (GFLNKGRP) is added in GF

Detailed Summary of GF Relationship to PF

Topic Variable	Values in Short Name of Genomic Measurement (GFTESTCD) and Name of Genomic Measurement (GFTEST) represent the genomic assessment as the topic for the record

Detailed Summary of GF Relationship to PF

Qualifier Variables	Values in Short Name of Genomic Measurement (GFTESTCD) and Name of Genomic Measurement (GFTEST) represent the genomic assessment as the topic for the record
	Measurement, Test, or Examination Detail (GFTSTDTL) represents the reportable from the genomic assessment
	Variables Category for Genomic Finding (GFCAT) and Subcategory for Genomic Finding (GFSCAT) are sponsor defined.

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

Qualifier Variables	Variable Inheritability (GFINHERT) represents whether the variation can be passed to the next generation. Mutation Type (PFMUTYP) is replaced
	New variable Genome Reference (GFGENREF) is added in GF
	New variable Chromosome (GFCHROM) is added in GF

Detailed Summary of GF Relationship to PF

Qualifier Variables	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
	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
	Variable Genetic Location of Interest (PFGENLI) is retired
	Variable Genetic Target (PFGENTRG) is retired

Detailed Summary of GF Relationship to PF

Qualifier Variables	Variable Sequence Identifier (GFSEQID) represents a unique identifier for the sequence used as the reference to identify the genetic variation in the result. Reference Sequence (PFREFSEQ) is replaced
	Variable Published Variant Identifier (GFPVRID) represents a unique identifier for the variation that has been publicly characterized in an external database. Reference SNP Cluster ID Number (PFRSNUM) is replaced
	Variable Copy Identifier (GFCOPYID) 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

Detailed Summary of GF Relationship to PF

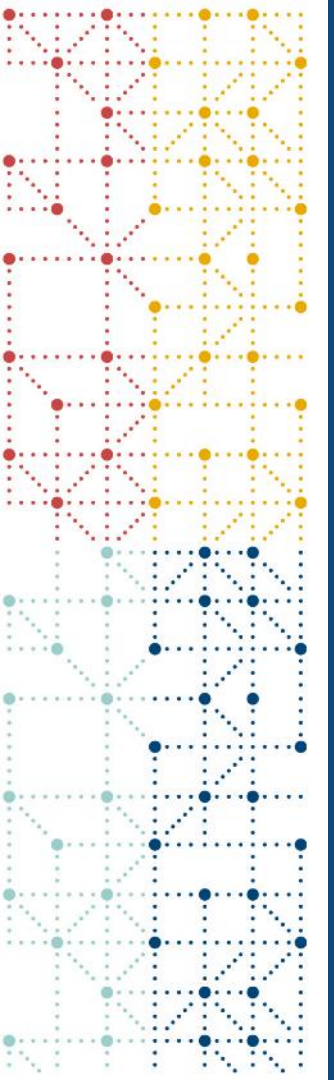
Qualifier Variables

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

Timing Variables	Established variable Time Point Reference (GFTPTREF) added in GF
	Established variable Date/Time of Reference Time Point (GFRFTDTC) added in GF



Questions & Answers

Audience Questions

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...)?

Audience Questions

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?

Audience Questions

If an organization has been using PF, 1) will it be required to and 2) is it straightforward to shift to using GF in it's 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.

Audience Questions

We would like more definition for GENLOC. Can you provide examples for cyogenetics where one or more chromosomes and related locations or location ranges?

Audience Questions

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?

Audience Questions

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

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Audience Questions

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?

Audience Questions

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?

Audience Questions

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!

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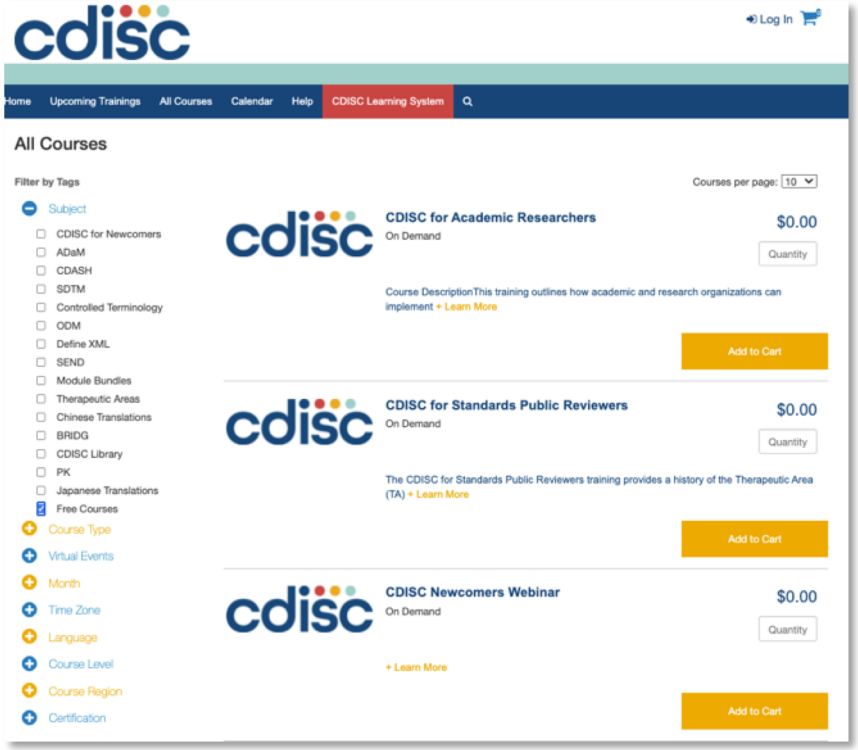

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Date	Webinars
24 March	Introduction to the SDTM Genomics Finding (GF) Domain
29 March	SDTM Office Hours
31 March	CDISC Open Source Alliance (COSA) Spotlight
5 April	Controlled Terminology Updates for Q1 – P49 Publication / P50 Public Review
19 April	QRS Office Hours
28 June	Controlled Terminology Updates for Q2 – P50 Publication / P51 Public Review

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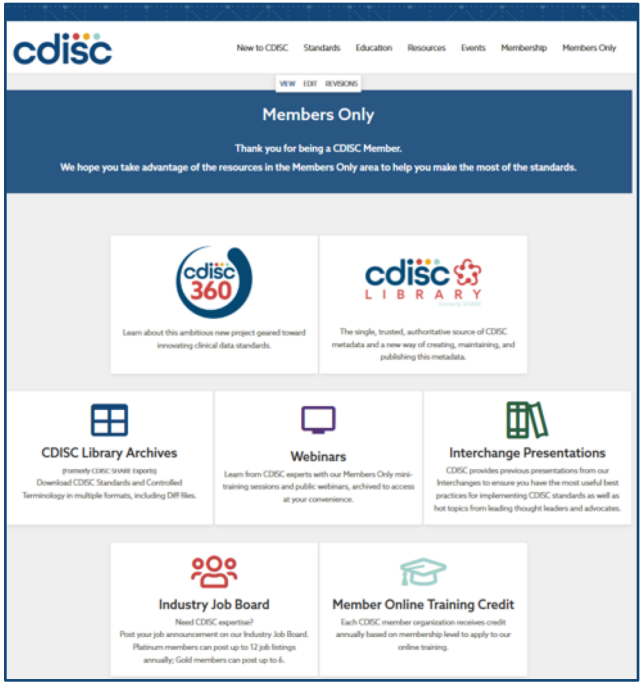

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