Leadership Team

- Christine Connolly
- Erin Muhlbradt
- Jon Neville

PGx Team

- See PGx Wiki Space

Terminology Team

- Erin Muhlbradt (NCI)
- Jordan Li (NCI)
- Chris Gemma (CDISC)

(Looking for more participants who are interested in designing domains for cytogenetics)

Team Mission

This project will support the submission of all types of genetic and genomics data including the use of any industry standards such as the International System for Human Cytogenetic Nomenclature (ISCN) and Human Genome Variation Society (HGVS).

Scope

The Pharmacogenomics/Genetics Implementation Guide (PGxIG) will be extended to include data structures to collect cytogenetic data with pertinent examples and terminology. We also intend to develop a PGx summary domain (PS) that represents the assay design. The team would also like to plan for Next Generation Sequencing and RNASEQ. Last year, the team published a Virology Guide focused on viral resistance that contained several examples for Viral Genetics. The current PGxIG effort, which should finish later this year includes extensions for mutations, SNP data and allelic results.

The team expects to perform on-going maintenance based on new data brought by team members to be mapped. The PGxIG team plans to provide an area in the CDISC wiki (Genomics Team Home) where we can develop and maintain a library of sample data that has been mapped to the domains.

The PGx team maintains its membership through careful selection of volunteer experts who can contribute to its various sub-teams with their technical or Therapeutic-based subject matter expertise.

2019 Work Plan

- Future Projects (Prioritized List)
  - Cytogenetics
  - CRF design for the collection of genetic/genomics data. (Joint with CDASH)
  - Gene Expression expansion to include population/cohort level reporting
  - Whole Genome Sequencing
  - Capturing Meta data that describes key aspects for trial design for genetic/genomic testing
  - Extend HLA examples for concepts such as typing ambiguity (allele and genotype lists) – when lab is not able to determine which alleles are present
  - Additional Allelic/Genotype uses cases
  - Miscellaneous Suggestions (from Public Review)

Stakeholders/Constituency

- Regulatory Authorities
- Standards Development Organizations
- Pharmaceutical Sponsors
- Medical Devices, Diagnostics
- Contract Research Organizations & Consultants

Dependencies & Collaborations

Dependencies on other Standards/Teams /SDOs /Organizations:
SDS team (SDTM), terminology, BRIDG, ODM/Define, SHARE, HL7 CG/LOINC and BRIDG

Collaborations:
SDS team (SDTM), Controlled Terminology, BRIDG, FDA, ODM/Define, SHARE, NCI-EVS, NCI-IRWG (BRIDG)