Date: February 2009

Phenotype and Genotype Object Model (PAGE-OM)

Beta 2 - with change bars

OMG Document Number: dtc/2009-02-03
Standard document URL: http://www.omg.org/spec/PAGE-OM/1.0/Beta2
Associated File(s)*: http://www.omg.org/spec/PAGE-OM/20080101
http://www.omg.org/spec/PAGE-OM/20080102
http://www.omg.org/spec/PAGE-OM/20080103

---

* original files: lifesci/2008-01-03 (PIM-normative), lifesci/2008-01-04 (PSM-normative), lifesci/2008-01-02 (normative and non-normative)

USE OF SPECIFICATION - TERMS, CONDITIONS & NOTICES

The material in this document details an Object Management Group specification in accordance with the terms, conditions and notices set forth below. This document does not represent a commitment to implement any portion of this specification in any company's products. The information contained in this document is subject to change without notice.

LICENSES

The companies listed above have granted to the Object Management Group, Inc. (OMG) a nonexclusive, royalty-free, paid up, worldwide license to copy and distribute this document and to modify this document and distribute copies of the modified version. Each of the copyright holders listed above has agreed that no person shall be deemed to have infringed the copyright in the included material of any such copyright holder by reason of having used the specification set forth herein or having conformed any computer software to the specification.

Subject to all of the terms and conditions below, the owners of the copyright in this specification hereby grant you a fully-paid up, non-exclusive, nontransferable, perpetual, worldwide license (without the right to sublicense), to use this specification to create and distribute software and special purpose specifications that are based upon this specification, and to use, copy, and distribute this specification as provided under the Copyright Act; provided that: (1) both the copyright notice identified above and this permission notice appear on any copies of this specification; (2) the use of the specifications is for informational purposes and will not be copied or posted on any network computer or broadcast in any media and will not be otherwise resold or transferred for commercial purposes; and (3) no modifications are made to this specification. This limited permission automatically terminates without notice if you breach any of these terms or conditions. Upon termination, you will destroy immediately any copies of the specifications in your possession or control.

PATENTS

The attention of adopters is directed to the possibility that compliance with or adoption of OMG specifications may require use of an invention covered by patent rights. OMG shall not be responsible for identifying patents for which a license may be required by any OMG specification, or for conducting legal inquiries into the legal validity or scope of those patents that are brought to its attention. OMG specifications are prospective and advisory only. Prospective users are responsible for protecting themselves against liability for infringement of patents.

GENERAL USE RESTRICTIONS

Any unauthorized use of this specification may violate copyright laws, trademark laws, and communications regulations and statutes. This document contains information which is protected by copyright. All Rights Reserved. No part of this work covered by copyright herein may be reproduced or used in any form or by any means--graphic, electronic, or mechanical, including photocopying, recording, taping, or information storage and retrieval systems--without permission of the copyright owner.

DISCLAIMER OF WARRANTY

WHILE THIS PUBLICATION IS BELIEVED TO BE ACCURATE, IT IS PROVIDED "AS IS" AND MAY CONTAIN ERRORS OR MISPRINTS. THE OBJECT MANAGEMENT GROUP AND THE COMPANIES LISTED ABOVE MAKE NO WARRANTY OF ANY KIND, EXPRESS OR IMPLIED, WITH REGARD TO THIS PUBLICATION, INCLUDING BUT NOT LIMITED TO ANY WARRANTY OF TITLE OR OWNERSHIP, IMPLIED WARRANTY OF MERCHANTABILITY OR WARRANTY OF FITNESS FOR A PARTICULAR PURPOSE OR USE.
IN NO EVENT SHALL THE OBJECT MANAGEMENT GROUP OR ANY OF THE COMPANIES LISTED ABOVE BE LIABLE FOR ERRORS CONTAINED HEREIN OR FOR DIRECT, INDIRECT, INCIDENTAL, SPECIAL, CONSEQUENTIAL, RELIANCE OR COVER DAMAGES, INCLUDING LOSS OF PROFITS, REVENUE, DATA OR USE, INCURRED BY ANY USER OR ANY THIRD PARTY IN CONNECTION WITH THE FURNISHING, PERFORMANCE, OR USE OF THIS MATERIAL, EVEN IF ADVISED OF THE POSSIBILITY OF SUCH DAMAGES.

The entire risk as to the quality and performance of software developed using this specification is borne by you. This disclaimer of warranty constitutes an essential part of the license granted to you to use this specification.

RESTRICTED RIGHTS LEGEND

Use, duplication or disclosure by the U.S. Government is subject to the restrictions set forth in subparagraph (c) (1) (ii) of The Rights in Technical Data and Computer Software Clause at DFARS 252.227-7013 or in subparagraph (c)(1) and (2) of the Commercial Computer Software - Restricted Rights clauses at 48 C.F.R. 52.227-19 or as specified in 48 C.F.R. 227-7202-2 of the DoD F.A.R. Supplement and its successors, or as specified in 48 C.F.R. 12.212 of the Federal Acquisition Regulations and its successors, as applicable. The specification copyright owners are as indicated above and may be contacted through the Object Management Group, 140 Kendrick Street, Needham, MA 02494, U.S.A.

TRADEMARKS

MDA®, Model Driven Architecture®, UML®, UML Cube logo®, OMG Logo®, CORBA® and XMI® are registered trademarks of the Object Management Group, Inc., and Object Management Group™, OMG™ , Unified Modeling Language™, Model Driven Architecture Logo™, Model Driven Architecture Diagram™, CORBA logos™, XMI Logo™, CWM™, CWM Logo™, IIOP™ , MOF™ , OMG Interface Definition Language (IDL)™ , and OMG Systems Modeling Language (OMG SysML)™ are trademarks of the Object Management Group. All other products or company names mentioned are used for identification purposes only, and may be trademarks of their respective owners.

COMPLIANCE

The copyright holders listed above acknowledge that the Object Management Group (acting itself or through its designees) is and shall at all times be the sole entity that may authorize developers, suppliers and sellers of computer software to use certification marks, trademarks or other special designations to indicate compliance with these materials.

Software developed under the terms of this license may claim compliance or conformance with this specification if and only if the software compliance is of a nature fully matching the applicable compliance points as stated in the specification. Software developed only partially matching the applicable compliance points may claim only that the software was based on this specification, but may not claim compliance or conformance with this specification. In the event that testing suites are implemented or approved by Object Management Group, Inc., software developed using this

specification may claim compliance or conformance with the specification only if the software satisfactorily completes the testing suites.

OMG's Issue Reporting Procedure

All OMG specifications are subject to continuous review and improvement. As part of this process we encourage readers to report any ambiguities, inconsistencies, or inaccuracies they may find by completing the Issue Reporting Form listed on the main web page *http://www.omg.org*, under Documents, Report a Bug/Issue (http://www.omg.org/technology/agreement.htm).

Table of Contents

Preface

About the Object Management Group

OMG

Founded in 1989, the Object Management Group, Inc. (OMG) is an open membership, not-for-profit computer industry standards consortium that produces and maintains computer industry specifications for interoperable, portable and reusable enterprise applications in distributed, heterogeneous environments. Membership includes Information Technology vendors, end users, government agencies and academia.

OMG member companies write, adopt, and maintain its specifications following a mature, open process. OMG's specifications implement the Model Driven Architecture® (MDA®), maximizing ROI through a full-lifecycle approach to enterprise integration that covers multiple operating systems, programming languages, middleware and networking infrastructures, and software development environments. OMG's specifications include: UML® (Unified Modeling Language™); CORBA® (Common Object Request Broker Architecture); CWM™ (Common Warehouse Metamodel); and industry-specific standards for dozens of vertical markets.

More information on the OMG is available at *http://www.omg.org/*.

OMG Specifications

As noted, OMG specifications address middleware, modeling and vertical domain frameworks. A catalog of all OMG Specifications is available from the OMG website at:

http://www.omg.org/technology/documents/spec_catalog.htm

Specifications within the Catalog are organized by the following categories:

OMG Modeling Specifications

- UML
- MOF
- XMI
- CWM
- Profile specifications.

OMG Middleware Specifications

- CORBA/IIOP
- IDL/Language Mappings
- Specialized CORBA specifications
- CORBA Component Model (CCM).

Platform Specific Model and Interface Specifications

- CORBAservices

- CORBAfacilities
- OMG Domain specifications
- OMG Embedded Intelligence specifications
- OMG Security specifications.

All of OMG's formal specifications may be downloaded without charge from our website. (Products implementing OMG specifications are available from individual suppliers.) Copies of specifications, available in PostScript and PDF format, may be obtained from the Specifications Catalog cited above or by contacting the Object Management Group, Inc. (as of January 16, 2006) at:

OMG Headquarters
140 Kendrick Street
Building A, Suite 300
Needham, MA 02494
USA
Tel: +1-781-444-0404
Fax: +1-781-444-0320
Email: *pubs@omg.org*

Certain OMG specifications are also available as ISO standards. Please consult *http://www.iso.org*

Issues

The reader is encouraged to report any technical or editing issues/problems with this specification to *http://www.omg.org/technology/agreement.htm*.

1 Scope

Please see Chapter 6 - Introduction.

2 Conformance

The normative parts of this specification are:

- Platform independent model expressed in the attached XML file created according to XMI format rules, v2.1, using program Enterprise Architect, version 7.0.817.1, UML version 2.1.
- Platform specific model, representing an XML-based data exchange format, defined by an XML Schema definition.

Any implementation using or producing data exchange format defined by the Platform specific model defined by this specification is considered complying with this specification.

Any platform specific model that is derived from the platform independent model defined by this specification is also considered compliant.

If there is any inconsistency, or discrepancy between generality and specificity, between the platform independent and platform specific model, the platform specific model has precedence.

The normative parts are expressed in the accompanied files in a document whose number is given in Appendix A (or elsewhere in this document). Parts of these files may also appear in the explanatory text of this document. If they do and if there are some differences or discrepancies the contents of the normative accompanied files has precedence.

Regarding the use of ontology, it is not normative to use the ones listed in the “Ontology” section of this document, but it is highly suggested. The reason why this specification is not stricter about it is the reflection of the fact that ontology is a moving target in the bio-community and insisting on using only specific ones, may harm the usability of this specification.

3 Normative References

There are no normative references associated with this specification.

4 Terms and Definitions

Please see Annex C – Glossary.

5 Supporting Organizations

The following organizations have been involved in the process of developing, prototyping, and/or reviewing this specification. The authors thank them for participating and giving their valuable input.

- Institute for Molecular Medicine Finland, University of Helsinki, Finland
- University of Bergen, Norway
- Cold Spring Harbor Laboratory (CSHL), USA
- National Center for Biotechnology Information (NCBI), USA
- Stanford University, USA
- Yale University, USA
- University College London,
- Centre for Health Informatics and Multiprofessional Education, UK
- University of Leicester, UK
- South African National Bioinformatics Institute, South Africa
- School of Medicine, Tsinghua University, China
- Shanghai Center for Bioinformation Technology, China
- Shanghai Information Center for Life Sciences, China
- Institute of Genomics & Integrative Biology (IGIB), India
- Japan Science and Technology Agency (JST), Japan
- Medical Institute of Bioregulation, Kyushu University, Japan
- Mie University Life Science Research Center, Japan
- National Institute of Genetics (NIG), Japan
- RIKEN, Japan
- Tokyo Medical and Dental University (TMDU), Japan
- Tokyo Metropolitan Institute of Gerontology, Japan
- Graduate School of Medicine, University of Tokyo, Japan
- UNISYS, USA

5.1 Acknowledgements

The publishers of this document wish to express their appreciation to those listed below (in non-significant order) for their contributions of ideas and experience. Ultimately, the ideas expressed in this document are those of the authors and do not necessarily reflect the views or ideas of these individuals, nor does the inclusion of their names imply an endorsement of the final product.

Kimitoshi Naito, Akihiko Konagaya, Albert V. Smith, Anthony J. Brookes, David Fredman, Debasis Dash, Haseena Rajeevan, Heikki Lehväslaiho, Hideaki Sugawara, Hiroshi Mizushima, Juha Muilu, Jun Nakaya, Katsushi Tokunaga, Kei-Hoi Cheung, Kenshi Hayashi, Mark Woon, Masako Kuroda, Masashi Tanaka, Martin Senger, Matthew Darlison, Takeshi Tomiki, Toshio Kojima, Yasumasa Shigemoto, Yoshiji Yamada.

6 Introduction

Modern biology is striving to understand what factors generate inter-individual differences in structure, development, or behaviour within a species. Genetic factors ('genotypes') are undoubtedly of key importance in this equation, as are environmental conditions and even stochastic events, and much research is being done to elucidate how these things impact a range of normal and disease 'phenotypes' (i.e., the characteristics that can be observed). Progress in this area will ultimately lead to improved and increasingly personalised medical care, more productive agricultural and farming systems, and better solutions for environmental monitoring and control.

Many organisms are being explored and utilised with regards to deciphering genotype-phenotype relationships. On the genetic level, it is now possible to determine DNA sequences at extremely high-throughput, thereby revealing normal and pathogenic variation in and around genes, across individuals. On the level of the phenotype, an almost unlimited number of things could be influenced by genetic variants, and increasingly precise and standardised assays are being devised to measure and assess such things.

Both genetic and phenotype datasets need to be managed and combined to elucidate genotype-phenotype relationships. The genetic datasets have so far mostly comprised assays of specific sites of variation (creating 'genotypes'), which have recently been growing exponentially due to technical advances. A subsequent new era is emerging wherein large DNA stretches (and perhaps complete genetic complements) are being fully (re)sequenced in relevant individuals. Phenotype datasets, both in human and non-human model organisms, are also being scaled up via the investigation of thousands or tens of thousands of individuals. Indeed, study integration is a major feature of current research; for example, existing Genome-wide Association (GWA) data done on various different studies are being merged in the ENAGE (ref) program leading to a potential GWA resource of 80,000 samples. As a result of this rapid progress in the modes of both genetic and phenotype analysis, the amount of genotype-phenotype data being generated is increasing at an exponential rate. The potential for converting this raw 'data' into useful ‘knowledge’ is therefore immense, but achieving this effectively will require distinct studies to be inter-related, cross-validated, and compared. In short, there is now an urgent need to ensure that genotype-phenotype investigations are supported by technologies that make them interoperable. Key to achieving this is a robust reference model for these types of data, via which all relevant domain knowledge can be effectively integrated and disseminated. That is the precisely the motivation behind building the Phenotype And GEnotype Object Model (PAGE-OM).

Anticipated user communities for PAGE-OM include biomedical researchers, clinicians, people involved in teaching and training such groups, and information technologists working in support of these domains.

Given the modern data production systems now at their disposal, biomedical researchers are constantly facing challenges of genotype-phenotype data management (e.g., storage, retrieval, tracking, reformatting, merging) and this directly impacts their ability to effectively analyse, share, and report their various datasets. Clinicians - for whom genotype-phenotype relationships are only one of many things to be considered - require streamlined and standardised access to explanations about DNA sequence alternatives and how and in which situations they may impact their clinical work. All of these needs will be helped by the development of PAGE-OM, which specifies the rational and functional segmentation of genotype-phenotype information into sub-components that are operationally distinct (the high-level objects in the model) and lays out the natural inter-relationships between these components. Once biomedical researchers and clinicians come to be familiar with these logical components and their relationships, it will be far easier for both groups to process and make sense of genotype-phenotype information, both within their own fields and in the important task of communicating between their domains of work. For this reason, it is also highly desirable that teachers of their two professions become familiar with the object modelling concepts, in order that they may suitably educate the researchers and doctors of the future.

Information technologists working in the biomedical fields will, more than any other group, benefit from understanding PAGE-OM in depth. These individuals have the job of providing the universe of databases, data pipelines, analysis tools, search functionalities, and exchanges protocols in which genotype-phenotype information will exist and be processed for exploitation. In the absence of any data model for genotype-phenotype information, these workers would each have to devise their own data model. Not only would this constant re-invention of the wheel be highly wasteful of manpower, it is also likely that many of the solutions they come up with would be suboptimal - given the limited experience that any one IT individual or IT team would usually have. Instead, PAGE-OM has been designed and piloted by a consortium of expert participants from nearly 50 Institutions engaged in many diverse genotype-phenotype projects. The PAGE-OM therefore provides a first-version common language, well-documented entity list, and carefully considered array of entity-relationships that IT workers can take directly off the shelf, with confidence that it should need nothing more than small adjustments or additions to serve their own specific needs. Additionally, as more and more IT groups adopt PAGE-OM and base their systems upon it, those systems will naturally become more and more similar and able to inter-communicate. This is an absolute necessity if the totality of genotype-phenotype information is ever to be merged into a single virtual corpus, for holistic and optimal utilisation.

To enable PAGE-OM to achieve its goals, it is structured as several high level concepts - each of which can be used as a standalone model with its own object classes and relationships thereof. These concepts are: Marker, Assay, Sample, Genotype, Frequency, Phenotype, and Experiment. By way of example uses; a company providing DNA analysis kits might only need to use the Marker and Assay parts of this model. A genome variation database might use solely the Marker component, or perhaps the Assay, Sample, Genotype, and Frequency entities as well listing variant findings in population groups. A project involved in collecting and examining clinical samples might use only Sample and Phenotype parts, and if that team subsequently undertook DNA analysis it could extend their data systems by incorporating some or all of the remaining concepts in order to fully describe whatever genotype-phenotype relationships they might discover. Such flexibility is a necessary and innate characteristic of PAGE-OM, and that extends into the way we have matched the design principles and various classes of the model to those of other data standards initiates that cover other domains of biomedical research.

In conclusion, we believe PAGE-OM provides a timely, robust, and useful data model, sufficiently developed and tested to justify formal registration and deployment to the many and various communities engaged in genotype-phenotype data handling.

7 Platform Independent Model

The platform independent model is expressed as a UML model, UML version 2.1. The normative is its XMI representation, attached in the “Accompanied files” as the file PIM/ PAGE-OM_uml_2.1_xmi_2.1.xml. The XMI was generated by the tool Enterprise Architect (EA), version 7.0.817.1, without including the EA specific extensions.

The XMI representing the platform independent model includes, because of the technical reasons and because of lacking package names, also the classes of the SNP-PML specification (that are used by this specification).

The basic data types (e.g., string or dateTime) are those inspired by the data types from the XML Schema (XML Schema Part 2: Datatypes - http://www.w3.org/TR/xmlschema-2/), where their exact descriptions can be found.

The full details with class and attributes description are attached in a generated file PIM/PAGE-OM.rtf. This file can be used interchangeably with the XMI file because both were generated from a model created by the Enterprise Architect tool - whose main file is also attached as file PIM/PAGE-OM.EAP. Note, however, that the latter file may have some EA specific expressions and extensions that are not normative for this specification.

For convenience, the whole PAGE-OM specification can be seen at http://www.pageom.org/models/omg/v_1.0/.

7.1 Detailed Model Documentation

7.1.1 PAGE-OM

Type: Package

Package: Model

7.1.1.1 Page

Type: Class

Package: PAGE-OM

Issue 13023 - new top level XML Element

Document:
root element of page xml-schema

Connections

Connector	Source	Target
Association Source ->Destination	PAGE-OM::Page	Page::Algorithm
Association Source ->Destination	PAGE-OM::Page	SNP2::Heterozygosity
Association Source ->Destination	PAGE-OM::Page	PAGE::Location_on_plate
Association Source ->Destination	PAGE-OM::Page	PAGE::Melting_temperature

Association Source ->Destination	PAGE-OM::Page	PAGE::Multi-variation_assay
Association Source ->Destination	PAGE-OM::Page	PAGE::Observable_feature_category
Association Source ->Destination	PAGE-OM::Page	PAGE::Observation_method
Association Source ->Destination	PAGE-OM::Page	PAGE::Observed_value
Association Source ->Destination	PAGE-OM::Page	PAGE::Frequency_set
Association **Source ->Destination**	PAGE-OM::Page	PAGE::Genotype_[henotype_correlation_ experiment
Association Source ->Destination	PAGE-OM::Page	PAGE::Experiment_result
Association Source ->Destination	PAGE-OM::Page	PAGE::Plate
Association Source ->Destination	PAGE-OM::Page	PAGE::Run
Association Source ->Destination	PAGE-OM::Page	SNP2::Assayed_genomic_genotype
Association Source ->Destination	PAGE-OM::Page	SNP2::Functional_change
Association Source ->Destination	PAGE-OM::Page	SNP2::Gene_based_haplotype
Association Source ->Destination	PAGE-OM::Page	SNP2::Genic_variation
Association Source ->Destination	PAGE-OM::Page	SNP2::Genomic_allele
Association Source ->Destination	PAGE-OM::Page	SNP2::Genomic_allele_population_ frequency
Association Source ->Destination	PAGE-OM::Page	PAGE::Phenotype_feature
Association Source ->Destination	PAGE-OM::Page	PAGE::Association_study
Association Source ->Destination	PAGE-OM::Page	PAGE::Lifestyle_feature
Association Source ->Destination	PAGE-OM::Page	SNP2::Genotype_haplotype
Association Source ->Destination	PAGE-OM::Page	SNP2::Band_size
Association Source ->Destination	PAGE-OM::Page	PAGE::Conclusion
Association Source ->Destination	PAGE-OM::Page	PAGE::Environment_feature
Association Source ->Destination	PAGE-OM::Page	PAGE::Evidence
Association Source ->Destination	PAGE-OM::Page	PAGE::Hypothesis
Association Source ->Destination	PAGE-OM::Page	Person
Association Source ->Destination	PAGE-OM::Page	Cytogenetic_map

Association Source ->Destination	PAGE-OM::Page	Db_xref
Association Source ->Destination	PAGE-OM::Page	Exon
Association Source ->Destination	PAGE-OM::Page	Genetic_location
Association Source ->Destination	PAGE-OM::Page	Genetic_map
Association Source ->Destination	PAGE-OM::Page	Genomic_reference_allele
Association Source ->Destination	PAGE-OM::Page	Geographic_location
Association Source ->Destination	PAGE-OM::Page	SNP2::Genomic_gene_structure
Association Source ->Destination	PAGE-OM::Page	Organization
Association Source ->Destination	PAGE-OM::Page	CDS
Association Source ->Destination	PAGE-OM::Page	Reference_genomic_assembly
Association Source ->Destination	PAGE-OM::Page	Reference_genomic_landmark
Association Source ->Destination	PAGE-OM::Page	Subject_descriptor
Association Source ->Destination	PAGE-OM::Page	Taxon
Association Source ->Destination	PAGE-OM::Page	BREF::Bibliographic_reference
Association Source ->Destination	PAGE-OM::Page	BREF::Bibref_description
Association Source ->Destination	PAGE-OM::Page	BREF::Journal
Association Source ->Destination	PAGE-OM::Page	BREF::Service
Association Source ->Destination	PAGE-OM::Page	Oligo
Association Source ->Destination	PAGE-OM::Page	SNP2::Panel
Association Source ->Destination	PAGE-OM::Page	SNP2::Genomic_haplotype_population_frequency
Association Source ->Destination	PAGE-OM::Page	SNP2::Genomic_variation
Association Source ->Destination	PAGE-OM::Page	SNP2::Haplotype_block
Association Source ->Destination	PAGE-OM::Page	SNP2::Haplotype_derivation_methods
Association Source ->Destination	PAGE-OM::Page	SNP2::Haplotype_map
Association Source ->Destination	PAGE-OM::Page	SNP2::Individual
Association Source ->Destination	PAGE-OM::Page	SNP2::Latent_genotype
Association Source ->Destination	PAGE-OM::Page	Cytogenetic_location

Association Source ->Destination	PAGE-OM::Page	SNP2::Neighbor_variation
Association Source ->Destination	PAGE-OM::Page	Contributor
Association Source ->Destination	PAGE-OM::Page	SNP2::Reference_genomic_location
Association Source ->Destination	PAGE-OM::Page	SNP2::Structural_change
Association Source ->Destination	PAGE-OM::Page	SNP2::Transcription_change
Association Source ->Destination	PAGE-OM::Page	SNP2::Translation_change
Association Source ->Destination	PAGE-OM::Page	SNP2::Variation_assay
Association Source ->Destination	PAGE-OM::Page	Anatomic_location
Association Source ->Destination	PAGE-OM::Page	Annotation
Association Source ->Destination	PAGE-OM::Page	SNP2::Genomic_genotype_population_frequency
Association Source ->Destination	PAGE-OM::Page	SNP2::Molecular_sample
Association Source ->Destination	PAGE-OM::Page	BREF::Source

7.1.2 PAGE

Type: Package «XSDschema»

Package: PAGE-OM

Evidence and value - *(Logical diagram)*

Core (“simple”) string, arithmetic data types and object references are modeled here, in the Value model. The model is based on concept developed in Generation Challenge Program: http://pantheon.generationcp.org/demeter/Values.html.

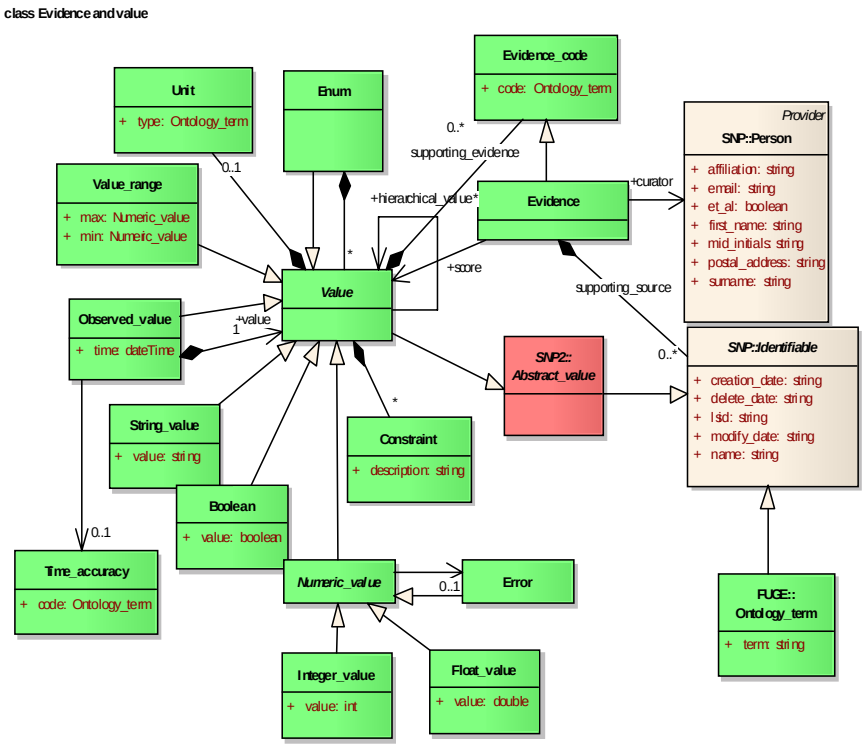

Figure 7.1

Issue 13004 - Value association renamed to hierarchical_value
13005 - XML schema element removed from diagrams
12998 - attribute “name” is removed

Frequency - (*Logical diagram*)

Alleles (Genomic_alleles), genotypes (Consensus_genomic_genotype) and haplotypes (Genomic_haplotype) can have measured frequencies in population samples (Panels).
In addition, heterozygosity (Heterozygosity) is a measure of observed variability of a polymorphic site (Genomic_variation) in a sub-population (Panel).

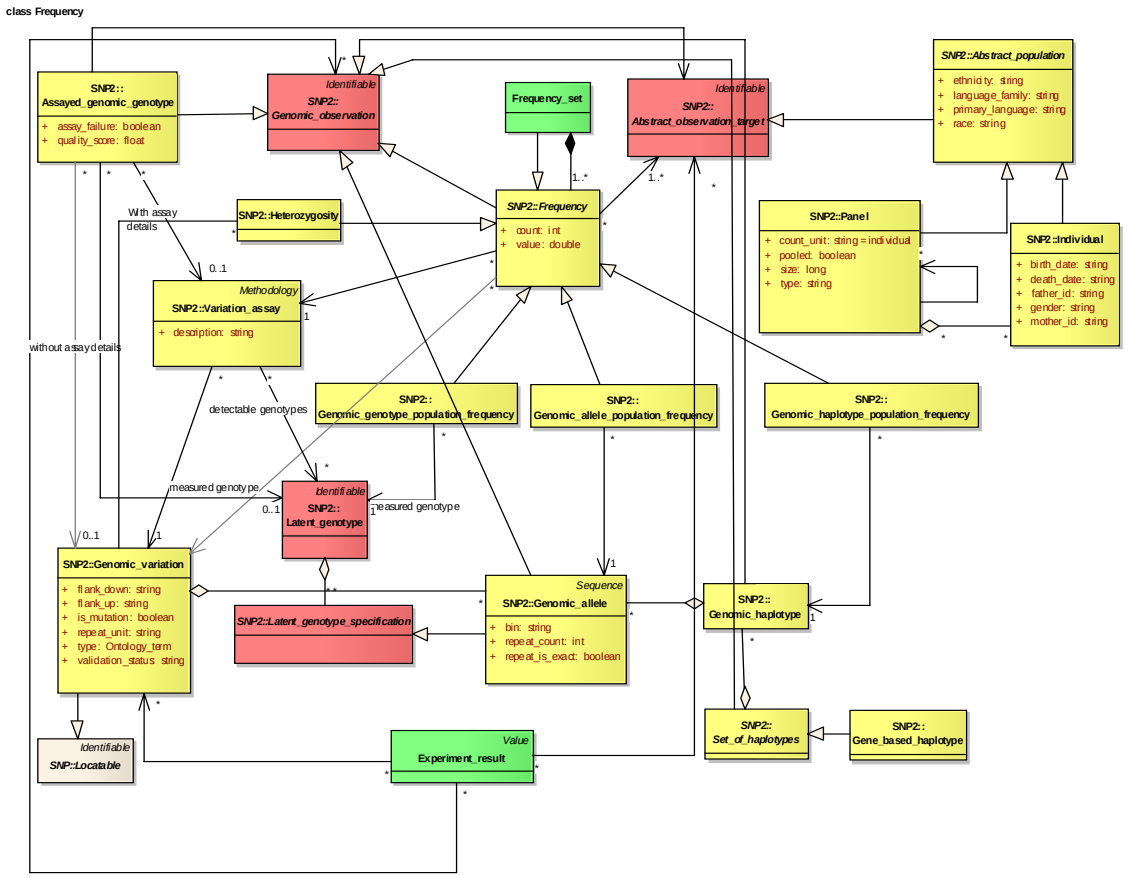

Figure 7.2

Issue 13008 - Attribute sex in Individual is renamed to gender

Genotype - (*Logical diagram*)

An assay (Variation_assay) is applied to a sample (Molecular_sample) to get a genotype.

(Assayed_genomic_genotype). One or more Assayed_genomic_genotype are used to reach a consensus (Consensus_genomic_genotype). The assays commonly use oligo primers to detect allelic variations (Genomic_alleles in Genomic_variation).

Assay can have reference to possible detectable latent genotypes (combination of alleles depending on ploidy). One of these combinations can be detected in single genotype measurement (Assayed_genomic_genotype) done using the specific assay.

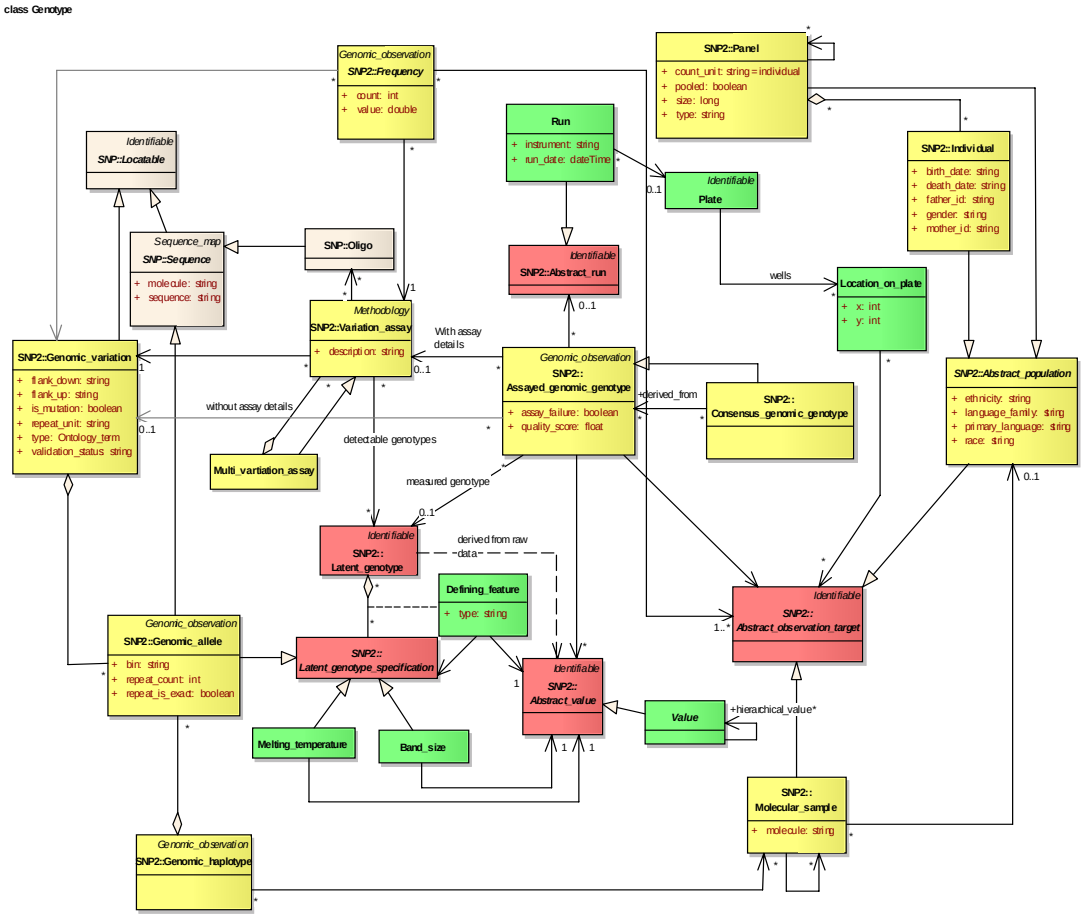

Figure 7.3

Issue 12999 - Attribute “derived from” renamed to “derived_from”
13001 - Aggregation from Consensus_geomic_genotype to Latent_genotype is deleted
13003 - Association Multivariation_assay to Genomic_variation is removed
13004 - Value association renamed to hierarchical_value
13007 - Frequency-Panel association moved to Frequency-Abstract_observation_target
13008 - Attribute sex in Individual is renamed to gender
13009 - Association Latent_
Genotype-Latent_genotype_specification changed to many-to-many

Genotype2 - (*Logical diagram*)

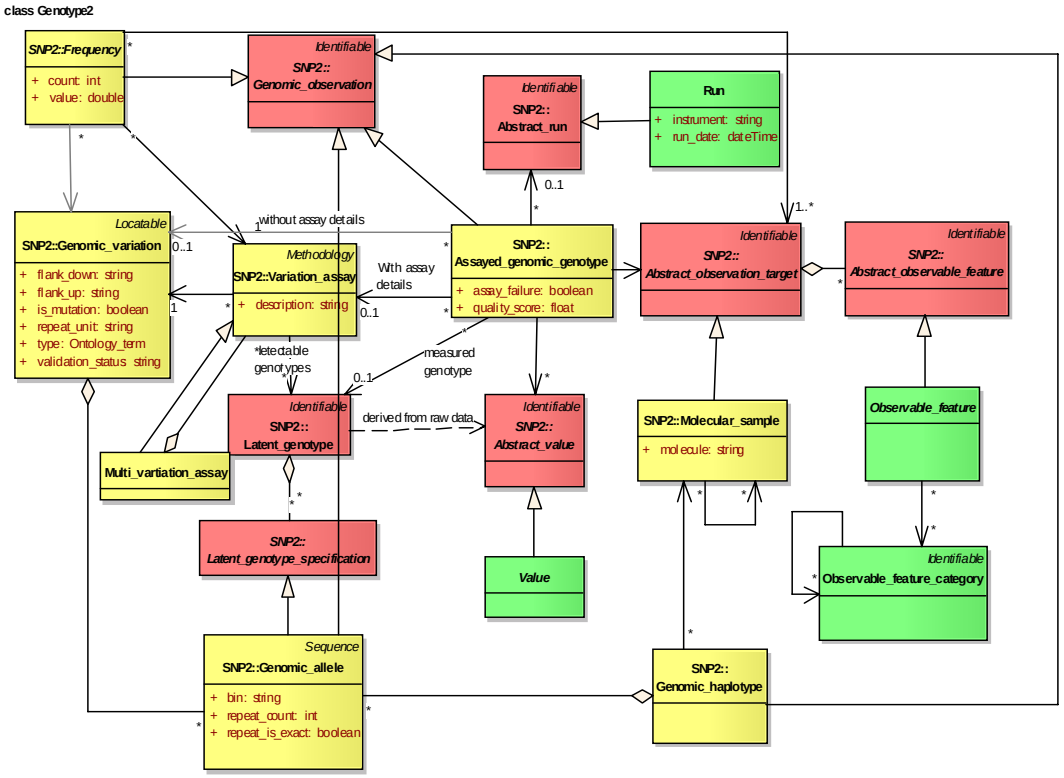

Figure 7.4

Issue 13003 - Association Multivariation_assay to Genomic_variation is removed
13007 - Frequency-Panel association moved to Frequency-Abstract_observation_target
13009 - Association Latent_
Genotype-Latent_genotype_specification changed to many-to-many
13010 - New Inheritance relationship: Genomic_allele is-a Genomic_observation

Identifiable - (*Logical diagram*)

Issue 13011 - Documentation of identifiable has changed

All classes in the model inherit from Identifiable. In this way, their instances are uniquely identifiable. Any Identifiable instance must use its “lsid attribute.” For this attribute, it is recommended to use the OMG Life Sciences Identifier specification. All classes in the model can be linked with annotation (Annotation) and database cross-reference (Db_xref). Special kinds of cross references are Source for source of data, Distributor for the original database of the data, and Contributor for tracking editorial changes to data.

Figure 7.5

Issue 13004 - Value association renamed to hierarchical_value

Phenotype - (*Logical diagram*)

Phenotype, environment and life style features are special cases of observable features from which observations can be made using specific observation methods. These observations lead to observed values obtained at specific time point from observation target (next diagram). Values can be also derived or categorized values (for example, high cholesterol level) in which case supporting values can be obtained from associated values, implemented using a recursion in the value model.

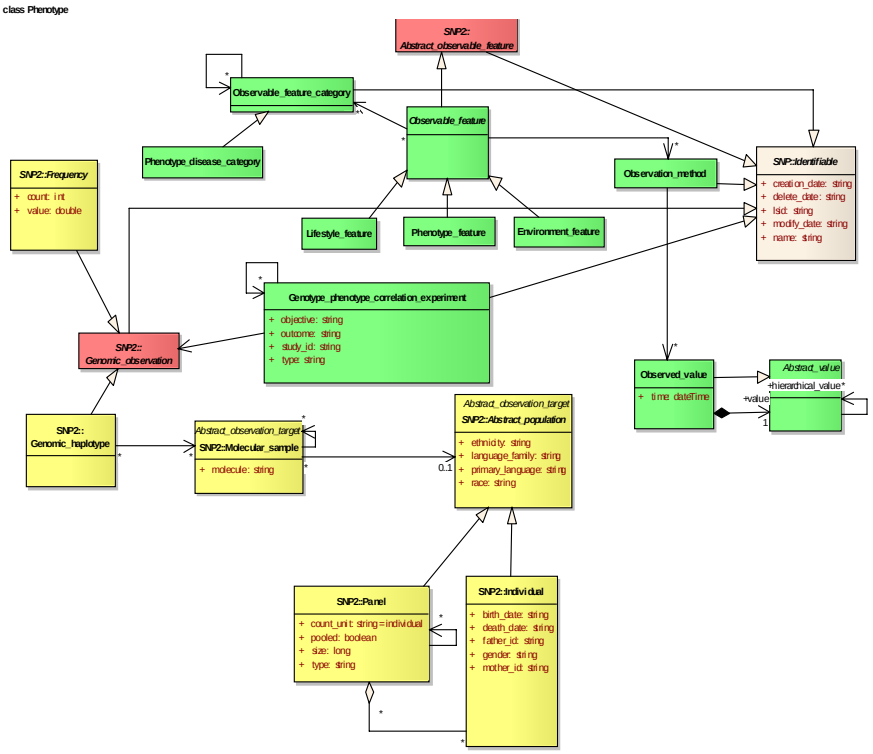

Figure 7.6

Issue 13004 - Value association renamed to hierarchical_value
13008 - Attribute sex in Individual is renamed to gender
13055 - Genotype_phenotype_correlation_experiment made non abstract

Phenotype 2 - (*Logical diagram*)

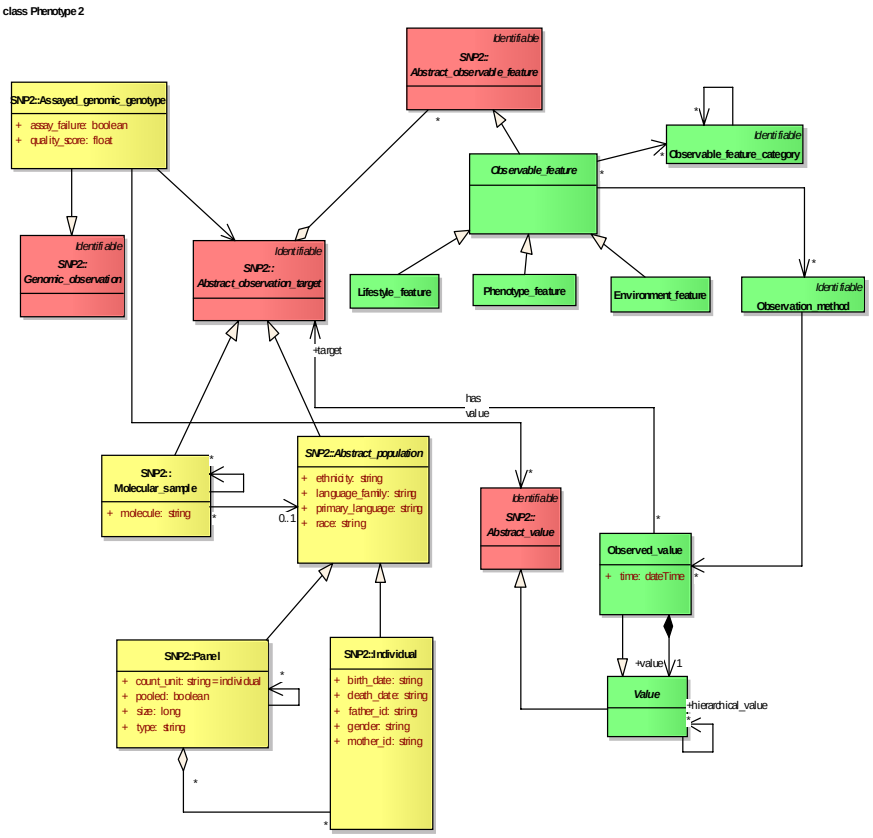

Figure 7.7

Issue 13004 - Value association renamed to hierarchical_value
13008 - Attribute sex in Individual is renamed to gender

Study - (*Logical diagram*)

Studies, like association studies, are composed from set of experiments done over observation targets. These experiments leads to set of results (Experiment_result) documented by genomic observations and observed phenotype values.

Figure 7.8

Issue 13004 - Value association renamed to hierarchical_value
13009 - Unsufficient cardinalities
Change cardinality of the association between Observed_value and Experiment_result from ‘many to 1’ to ‘many to many.’ Change cardinality of the association between Genotype_phenotype_correlation_experiment and Experiment_result from ‘1 to many’ to ‘many to many.’
13055 - Genotype_phenotype_correlation_experiment made non abstract

7.1.2.1 Algorithm

Type: Class_Identifiable

Package: PAGE

Document:

Step-by-step procedure for solving a problem.

Connections

Connector	Source	Target
Association Source -> Destination	PAGE-OM::Page	PAGE::Algorithm
Generalization Source -> Destination	PAGE::Algorithm	Identifiable
Association Source -> Destination	PAGE::Experiment_result	PAGE::Algorithm

Attributes

Attribute	Notes
description string	Description of algorithm

7.1.2.2 Association_study

Type: Class_Study

Package: PAGE

Document:

Examination of genetic variation across the human genome, designed to identify genetic associations with observable phenotypes.

Connections

Connector	Source	Target
Association Source -> Destination	PAGE-OM::Page	PAGE::Association_study
Generalization Source -> Destination	PAGE::Association_study	PAGE::Study
Aggregation Source -> Destination	PAGE::Genotype_phenotype_correlation_experiment	PAGE::Association_study

7.1.2.3 Band_size

Type: Class_Latent_genotype_specification

Package: PAGE

Document:

DNA fragment length estimated from gel electrophoresis

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Band_size	SNP2::Latent_genotype_specification
Association Source -> Destination	PAGE-OM::Page	PAGE::Band_size
Association Source -> Destination	PAGE::Band_size	SNP2::Abstract_value

7.1.2.4 Boolean

Type: Class_Value

Package: PAGE

Document:

Value of type boolean

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Boolean	PAGE::Value

Attributes

Attribute	Notes
value boolean	Boolean value

7.1.2.5 Conclusion

Type: Class_Identifiable

Package: PAGE

Document:

A reasoned judgment of an experiment

Connections

Connector	Source	Target
Association Source -> Destination	PAGE::Genotype_phenotype_correlation_experiment	PAGE::Conclusion
Association Source -> Destination	PAGE-OM::Page	PAGE::Conclusion
Generalization Source -> Destination	PAGE::Conclusion	Identifiable

Attributes

Attribute	Notes
p_value double	Probability value

7.1.2.6 Constraint

Type: Class

Package: PAGE

Document:

The contents of a Value can be limited by Constraints. Different types of Constraints allow various ways how to limit or validate one or more Value instances. The Constraint superclass only stores a string description of the Constraint. The actual full semantics of a constraint are specified in various subclasses described below.

But there are no subclasses in the PAGE-OM - because they are out of scope of PAGE-OM.

Connections

Connector	Source	Target
Aggregation Source -> Destination	PAGE::Constraint	PAGE::Value

Attributes

Attribute	Notes
description string	Description

7.1.2.7 Defining_feature

Type: AssociationClass

Package: PAGE

Document:

Association class has list of values which are used in defining the instance of Latent_genotype_specifications (for example intensity values used in allele calling).

Connections

Connector	Source	Target
Association Source -> Destination	PAGE::Defining_feature	SNP2::Abstract_value
Association Source -> Destination	PAGE::Defining_feature	SNP2::Latent_genotype_specification

Attributes

Attribute	Notes
type string	Type of feature

7.1.2.8 Enum

Type: Class_Value

Package: PAGE

Document:

Enumeration contains list of Values

Connections

Connector	Source	Target
Aggregation Source -> Destination	PAGE::Value	PAGE::Enum
Generalization Source -> Destination	PAGE::Enum	PAGE::Value

7.1.2.9 Environment_feature

Type: Class_Observable_feature

Package: PAGE

Document:

Circumstances, objects, or conditions by which one is surrounded.

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Environment_feature	PAGE::Observable_feature
Association Source -> Destination	PAGE-OM::Page	PAGE::Environment_feature

7.1.2.10 Error

Type: Class_Numeric_value

Package: PAGE

Document:

Error value is numeric value of accuracy. Quality score

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Error	PAGE::Numeric_value
Association Source -> Destination	PAGE::Numeric_value	PAGE::Error

7.1.2.11 Evidence

Type: Class_Evidence_code

Package: PAGE

Document:

Evidence indicates reliability of a feature or simply documents its authoritative origin.

Connections

Connector	Source	Target
Aggregation supporting_source Source -> Destination	Identifiable	PAGE::Evidence
Association Source -> Destination	PAGE::Evidence	score score of value PAGE::Value
Association Source -> Destination	PAGE-OM::Page	PAGE::Evidence
Generalization Source -> Destination	PAGE::Evidence	PAGE::Evidence_code
Association Source -> Destination	PAGE::Evidence	curator Curator of evidence Person

7.1.2.12 Evidence_code

Type: Class

Package: PAGE

Document:

Evidence can be an EvidenceCode (which is a controlled vocabulary term such as a GO evidence code or ICIS Method code) but can be a more fully documented Evidence object (inheriting from EvidenceCode) generally curated by a specified person, a curator modeled as a Contact.

Its strength is expressed by the score (which is usually a numeric value between 0 and 1, but also other types of Value are allowed - e.g., an ontology term value).

The core of an evidence is its supporting source which can be anything (because it is identified by a SimpleIdentifier). Usual evidence sources are BiblioReferences, Studies and OntologyTerms.Reference (generationcp - http://pantheon.generationcp.org/demeter/Features.html).

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Evidence	PAGE::Evidence_code
Aggregation supporting_evidence Source -> Destination	PAGE::Evidence_code	PAGE::Value

Attributes

Attribute	Notes
code Ontology_term	Evidence code as specified using ontology term.

7.1.2.13 Experiment_result

Type: Class_Value

Package: PAGE

Document:

Result of experiment

Connections

Connector	Source	Target
Association Source -> Destination	PAGE-OM::Page	PAGE::Experiment_result
Association Source -> Destination	PAGE::Experiment_result	SNP2::Genomic_observation
Association Result Source -> Destination	PAGE::Genotype_phenotype_correlation_experiment	PAGE::Experiment_result
Association Source -> Destination	PAGE::Experiment_result	PAGE::Algorithm
Generalization Source -> Destination	PAGE::Experiment_result	PAGE::Value
Association Source -> Destination	PAGE::Experiment_result	SNP2::Abstract_observation_target
Association Source -> Destination	PAGE::Experiment_result	PAGE::Observed_value
Association Source -> Destination	PAGE::Experiment_result	SNP2::Genomic_variation

7.1.2.14 Float_value

Type: Class_Numeric_value

Package: PAGE

Document:

Value of type float

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Float_value	PAGE::Numeric_value

Attributes

Attribute	Notes
value double	Value

7.1.2.15 Frequency_set

Type: Class_Frequency

Package: PAGE

Document:

Set of frequencies

Connections

Connector	Source	Target
Association Source -> Destination	PAGE-OM::Page	PAGE::Frequency_set
Generalization Source -> Destination	PAGE::Frequency_set	SNP2::Frequency
Aggregation Source -> Destination	SNP2::Frequency	PAGE::Frequency_set

7.1.2.16 Genotype_phenotype_correlation_experiment

Issue 13055 - Genotype_phenotype_correlation_experiment made non abstract

Type: Class_Identifiable

Package: PAGE

Document:

Family or case control based association study

Connections

Connector	Source	Target
Association Source -> Destination	PAGE::Genotype_phenotype_correlation_ experiment	PAGE::Genotype_phenotype_correlation_ experiment
Association Source -> Destination	PAGE::Genotype_phenotype_correlation_ experiment	PAGE::Conclusion
Association Source -> Destination	PAGE-OM::Page	PAGE::Genotype_phenotype_correlation_ experiment
Association Result Source -> Destination	PAGE::Genotype_phenotype_correlation_ experiment	PAGE::Experiment_result
Association Source -> Destination	PAGE::Genotype_phenotype_correlation_ experiment	SNP2::Genomic_observation
Association Observation targets used in experiment Source -> Destination	PAGE::Genotype_phenotype_correlation_ experiment	SNP2::Abstract_observation_target
Aggregation Source -> Destination	PAGE::Genotype_phenotype_correlation_ experiment	PAGE::Association_study
Association Source -> Destination	PAGE::Genotype_phenotype_correlation_ experiment	PAGE::Hypothesis
Association Genomic variations used in experiment Source -> Destination	PAGE::Genotype_phenotype_correlation_ experiment	SNP2::Genomic_variation
Generalization Source -> Destination	PAGE::Genotype_phenotype_correlation_ experiment	Identifiable

Attributes

Attribute	Notes
objective string	Objective of experiment
outcome string	Outcome of experiment
study_id string	Identifier of study
type string	Type of experiment

7.1.2.17 Hypothesis

Type: Class_Identifiable

Package: PAGE

Document:

Hypothesis of study

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Hypothesis	Identifiable
Association Source -> Destination	PAGE-OM::Page	PAGE::Hypothesis
Association Source -> Destination	PAGE::Genotype_phenotype_correlation_experiment	PAGE::Hypothesis

Attributes

Attribute	Notes
description string	Description of hypothesis

7.1.2.18 Integer_value

Type: Class_Numeric_value

Package: PAGE

Document:

Value of type integer

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Integer_value	PAGE::Numeric_value

Attributes

Attribute	Notes
value int	Integer value

7.1.2.19 Lifestyle_feature

Type: Class_Observable_feature

Package: PAGE

Document:

Way of life of an individual or panel

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Lifestyle_feature	PAGE::Observable_feature
Association Source -> Destination	PAGE-OM::Page	PAGE::Lifestyle_feature

7.1.2.20 Location_on_plate

Type: Class

Package: PAGE

Document:

X,Y plate_positions (wells). Numbering starts from one. Each well can contain one or more observation targets (molecular samples) prepared for measurement using one or more variation assays (e.g., assay multiplexing Assay_set). **Note**: These are optional laboratory specific details (Sample and Assay information is in Assayed_genomic_genotype).

Connections

Connector	Source	Target
Association wells Source -> Destination	PAGE::Plate	PAGE::Location_on_plate
Association Source -> Destination	PAGE::Location_on_plate	SNP2::Abstract_observation_target
Association Source -> Destination	PAGE-OM::Page	PAGE::Location_on_plate

Attributes

Attribute	Notes
x int	x coordinate of plate.
y int	y coordinate of plate

7.1.2.21 Melting_temperature

Type: Class_Latent_genotype_specification

Package: PAGE

Document:

The temperature at which DNA goes from a double-stranded to a single-stranded state. Unit of temperature is Celsius.

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Melting_temperature	SNP2::Latent_genotype_specification
Association Source -> Destination	PAGE-OM::Page	PAGE::Melting_temperature
Association Source -> Destination	PAGE::Melting_temperature	SNP2::Abstract_value

7.1.2.22 Multi_vartiation_assay

Type: Class_Variation_assay

Package: PAGE

Document:

Multi_variation_assay is a collection of assays which may be used simultaneously. Examples would be multiplex assays, micro-array based assays, or a panel of single-plex assays that share some common feature or purpose.

Issue 13003 - Association Multi-variation_assay to Genomic_variation is removed

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Multi_vartiation_assay	SNP2::Variation_assay
Aggregation Source -> Destination	SNP2::Variation_assay	PAGE::Multi_vartiation_assay
Association Source -> Destination	PAGE-OM::Page	PAGE::Multi_vartiation_assay

7.1.2.23 Numeric_value

Type: Class_Value

Package: PAGE

Document:

Numeric value

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Integer_value	PAGE::Numeric_value
Generalization Source -> Destination	PAGE::Float_value	PAGE::Numeric_value
Generalization Source -> Destination	PAGE::Error	PAGE::Numeric_value
Association Source -> Destination	PAGE::Numeric_value	PAGE::Error
Generalization Source -> Destination	PAGE::Numeric_value	PAGE::Value

7.1.2.24 Observable_feature

Type: Class_Abstract_observable_feature

Package: PAGE

Document:

Measurable feature of observable (e.g., size of nose).

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Observable_feature	SNP2::Abstract_observable_feature
Generalization Source -> Destination	PAGE::Environment_feature	PAGE::Observable_feature
Association Source -> Destination	PAGE::Observable_feature	PAGE::Observable_feature_category
Association Source -> Destination	PAGE::Observable_feature	PAGE::Observation_method
Generalization Source -> Destination	PAGE::Phenotype_feature	PAGE::Observable_feature
Generalization Source -> Destination	PAGE::Lifestyle_feature	PAGE::Observable_feature

7.1.2.25 Observable_feature_category

Type: Class_Identifiable

Package: PAGE

Document:

Phenotype category or ontology

Connections

Connector	Source	Target
Association Source -> Destination	PAGE::Observable_feature_category	PAGE::Observable_feature_category
Generalization Source -> Destination	PAGE::Phenotype_disease_category	PAGE::Observable_feature_category

Association Source -> Destination	PAGE::Observable_feature	PAGE::Observable_feature_category
Association Source -> Destination	PAGE-OM::Page	PAGE::Observable_feature_category
Generalization Source -> Destination	PAGE::Observable_feature_category	Identifiable

7.1.2.26 Observation_method

Type: Class_Identifiable

Package: PAGE

Document:

Method of observation (e.g., ruler). Can be also questionnaire.

Connections

Connector	Source	Target
Association Source -> Destination	PAGE::Observable_feature	PAGE::Observation_method
Association Source -> Destination	PAGE-OM::Page	PAGE::Observation_method
Association Source -> Destination	PAGE::Observation_method	PAGE::Observed_value
Generalization Source -> Destination	PAGE::Observation_method	Identifiable

7.1.2.27 Observed_value

Type: Class_Value

Package: PAGE

Document:

Observation done at specific point in time.

Connections

Connector	Source	Target
Association Source -> Destination	PAGE::Observed_value	PAGE::Time_accuracy
Association Source -> Destination	PAGE-OM::Page	PAGE::Observed_value
Association has value Source -> Destination	PAGE::Observed_value	target SNP2::Abstract_observation_target
Association Source -> Destination	PAGE::Observed_value	value PAGE::Value
Generalization Source -> Destination	PAGE::Observed_value	PAGE::Value
Association Source -> Destination	PAGE::Observation_method	PAGE::Observed_value
Association Source -> Destination	PAGE::Experiment_result	PAGE::Observed_value

Attributes

Attribute	Notes
time dateTime	Time of observation

7.1.2.28 Phenotype_disease_category

Type: Class_Observable_feature_category

Package: PAGE

Document:

Disease ontology.

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Phenotype_disease_category	PAGE::Observable_feature_category

7.1.2.29 Phenotype_feature

Type: Class_Observable_feature

Package: PAGE

Document:

Observable part of the structure, function, or behavior of a living organism.

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Phenotype_feature	PAGE::Observable_feature
Association Source -> Destination	PAGE-OM::Page	PAGE::Phenotype_feature

7.1.2.30 Plate

Type: Class_Identifiable

Package: PAGE

Document:

A sample holder, for example a microtiter plate used in one or many Runs. Samples (Observation_targets) are positioned on the plate using Location_on_plate-class.

Connections

Connector	Source	Target
Association wells Source -> Destination	PAGE::Plate	PAGE::Location_on_plate
Generalization Source -> Destination	PAGE::Plate	Identifiable
Association Source -> Destination	PAGE::Run	PAGE::Plate
Association Source -> Destination	PAGE-OM::Page	PAGE::Plate

7.1.2.31 Run

Type: Class_Abstract_run

Package: PAGE

Document:

The class contains information on execution of measurement experiments like time of execution and name of instrument.

Connections

Connector	Source	Target
Association Source -> Destination	PAGE::Run	PAGE::Plate
Association Source -> Destination	PAGE-OM::Page	PAGE::Run
Generalization Source -> Destination	PAGE::Run	SNP2::Abstract_run

Attributes

Attribute	Notes
instrument string	Name of the instrument
run_date dateTime	Date of run.

7.1.2.32 String_value

Type: Class_Value

Package: PAGE

Document:

Value of type string

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::String_value	PAGE::Value

Attributes

Attribute	Notes
value string	Actual value

7.1.2.33 Study

Type: Class_Identifiable

Package: PAGE

Document:

Research study

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Association_study	PAGE::Study
Generalization Source -> Destination	PAGE::Study	Identifiable

Attributes

Attribute	Notes
abstract string	Abstract
acknowledgements string	Acknowledgements
background string	Background information
conclusions string	Conclusions
key_results string	Key findings
limitations string	Limitations
objectives string	Objectives of study
source_of_bias string	Possible source of bias
study_design string	Study design
study_power string	Power of study
study_size_reason string	Reason for study size
submission_date date	Submission date of study
title string	Title of study
update_date date	Date when study is updated

7.1.2.34 Time_accuracy

Type: Class

Package: PAGE

Document:

Accuracy code for time. Accuracy code contains information on incompleteness of time. For example in some cases exact time is not known or cannot be given accurately for various reasons.

Connections

Connector	Source	Target
Association Source -> Destination	PAGE::Observed_value	PAGE::Time_accuracy

Attributes

Attribute	Notes
code Ontology_term	Accuracy code as defined in specific ontology.

7.1.2.35 Unit

Type: Class

Package: PAGE

Document:

Unit of value. Unit is defined using ontology term.

Connections

Connector	Source	Target
Aggregation Source -> Destination	PAGE::Unit	PAGE::Value

Attributes

Attribute	Notes
type Ontology_term	Type of unit

7.1.2.36 Value

Type: Class_Abstract_value

Package: PAGE

Document:

Abstract class. Extension point for Value implementations. Value model is based on concept developed in Generation Challenge Program: http://pantheon.generationcp.org/demeter/Values.html.

Issue 13004 - Value association renamed to hierarchical_value

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Value	SNP2::Abstract_value
Aggregation Source -> Destination	PAGE::Value	PAGE::Enum
Generalization Source -> Destination	PAGE::Value_range	PAGE::Value
Generalization Source -> Destination	PAGE::String_value	PAGE::Value
Generalization Source -> Destination	PAGE::Boolean	PAGE::Value
Aggregation Source -> Destination	PAGE::Constraint	PAGE::Value
Association Source -> Destination	PAGE::Evidence	score score of value PAGE::Value
Generalization Source -> Destination	PAGE::Experiment_result	PAGE::Value
Association Source -> Destination	PAGE::Observed_value	value PAGE::Value
Generalization Source -> Destination	PAGE::Observed_value	PAGE::Value
Aggregation Source -> Destination	PAGE::Unit	PAGE::Value

Generalization Source -> Destination	PAGE::Enum	PAGE::Value
Association Source -> Destination	PAGE::Value	hierarchical_value PAGE::Value
Generalization Source -> Destination	PAGE::Numeric_value	PAGE::Value
Aggregation supporting_evidence Source -> Destination	PAGE::Evidence_code	PAGE::Value

7.1.2.37 Value_range

Type: Class_Value

Package: PAGE

Document:

Inclusive value range

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Value_range	PAGE::Value

Attributes

Attribute	Notes
max Numeric_value	maximum value
min Numeric_value	minimum value

7.1.3 SNP2

Type: Package «XSDschema»

Package: PAGE-OM

Haplotype - *(Logical diagram)*

Haplotype (Genomic_haplotype) is a set of polymorphisms (Genomic alleles) on a single chromosome (chromatid). Haplotypes may be grouped into sets like haplotype blocks (Haplotype_block) separated by recombination regions and gene based haplotypes (Gene_based_haplotype), which combines sets of haplotypes on same gene structure.

Depending on an assay used, a genotype (Consensus_genomic_genotype) may contain information about the phase of the detected Genomic_alleles. Alternatively, various additional methods (Haplotype_derivation_methods) can be used to measure or calculate haplotypes (Genomic_haplotypes) from the genotype data.

The aim of many haplotype studies is to find haplotypes within a given sequence region that define most of the variation in populations

(Set_of_haplotypes). Haplotype blocks

(Haplotype_blocks) are clusters determined by linkage disequilibrium-based methods. The other common way to define clusters is use a the sequence region of the gene

(Gene_based_haplotype).

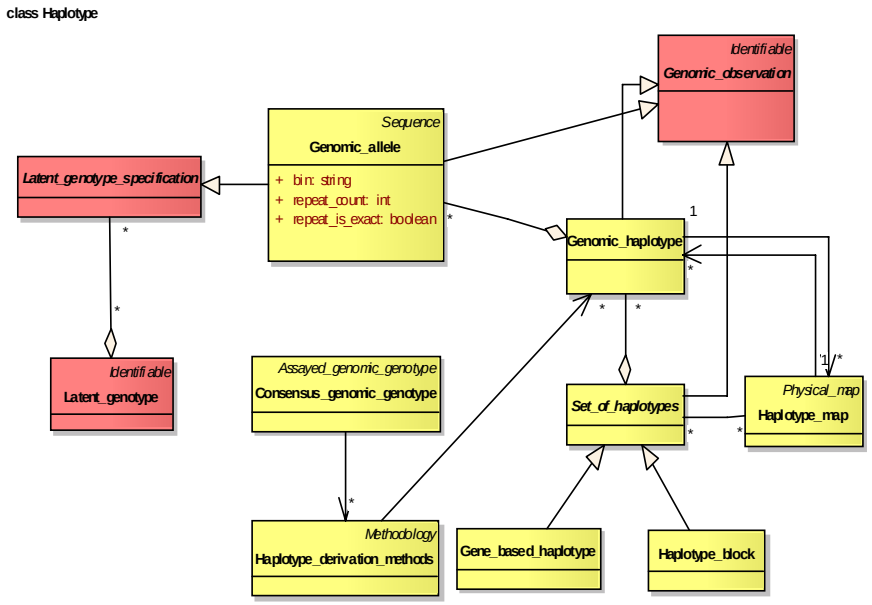

Figure 7.9

Issue 13002 - Redundant inheritance from Identifiable is removed in Set_of_haplotypes
13010 - New Inheritance relationship: Genomic_allele is-a Genomic_observation

Map - (Logical diagram)

A map organizes genomic features and assigns them locations. The primary maps (Physical_map, Sequence_map), which have additive sequence residue count distances between markers, are separated here from secondary maps (Ordered_map) that at best can only deal with non-metric distances. For each type a map (Map) there is a corresponding location (Location). Hence a marker (Genetic_variation) can have several locations (Reference_genomic_location) in sequences (Reference_genomic_landmark) and additionally be located within a band (Cytogenetic_location) in a species-specific karyotype (Cytogenetic_map) as well as in (Genetic_location) several genetic maps (Genetic_map), e.g., in maps based on male and female recombination frequencies. Genomic assembly (Reference_genomic_assembly) is a physical sequence map that is combination of genomic sequences (Reference_genomic_landmark). Feature locations can also be expressed in chromosomal locations in an assembly(Reference_genomic_location_in_assembly).

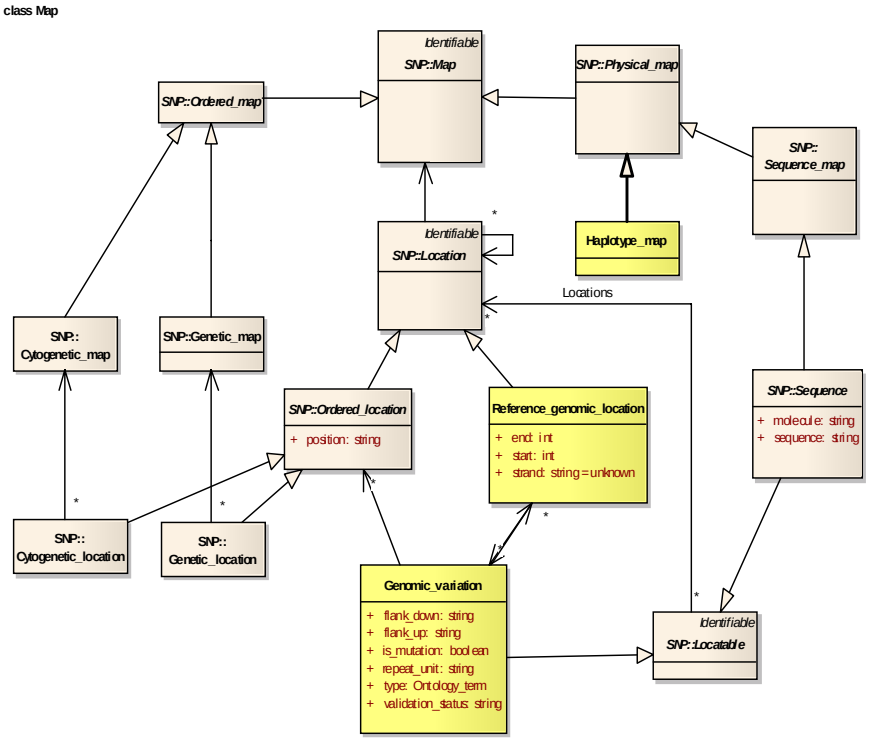

Figure 7.10

Issue 12998 - attribute “name” is removed

Sample - (Logical diagram)

An Individual (Individual) interacts with the rest of the model by giving one or more samples (Molecular_samples) from different tissues. Mixed samples and more complicated sample organisations are modeled by allowing Molecular_samples and Panels be composed of multiples of themselves. In these cases it is common that individuals can not be identified.

An individual (Individual) has parent IDs as attributes. This information makes it possible to reconstruct pedigrees when needed. A taxon identifies the taxonomic group, typically species, the individual or the population sample belongs to. Further, they can be place on a geographic map (Geographic_location).

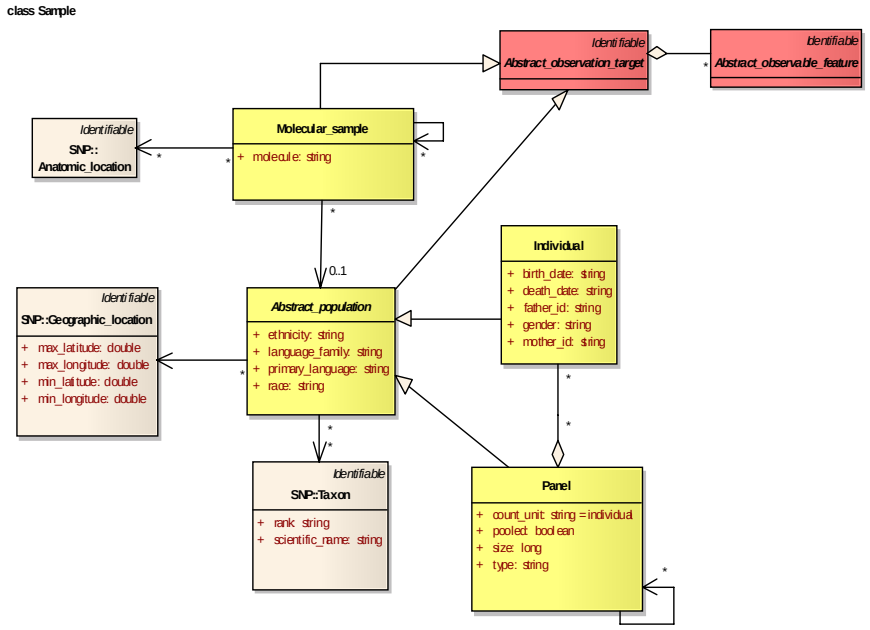

Figure 7.11

Sequence - (Logical diagram)

All sequences inherit from a generic Sequence class. All sequences (Sequence) and sequence features (e.g., Genomic_variation) can be located within a genomic sequence (Reference_genomic_sequence). This is modeled by common inheritance from an abstract superclass Locatable that can have several locations (Genomic_reference_location).

A Reference_genomic_landmark is any accessioned sequence within Reference_genomic_assembly. The model allows for multiple assemblies. Any location within a landmark and therefore in an assembly is called Reference_genomic_location. Any variable site in an assembly is a Genomic_polymorphism. The variable sequences within Genomic_variations are called Genomic_alleles.

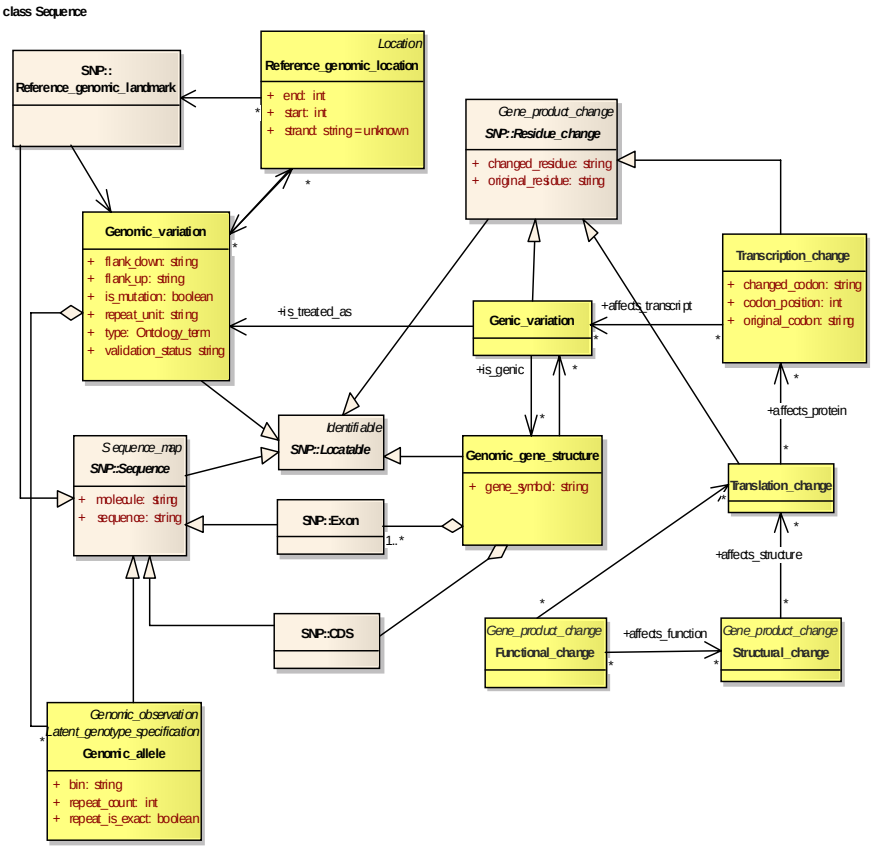

Figure 7.12

Issue 12999 - Attribute “is treated as” renamed to “is_treated_as”

Sequence 2 - (*Logical diagram*)

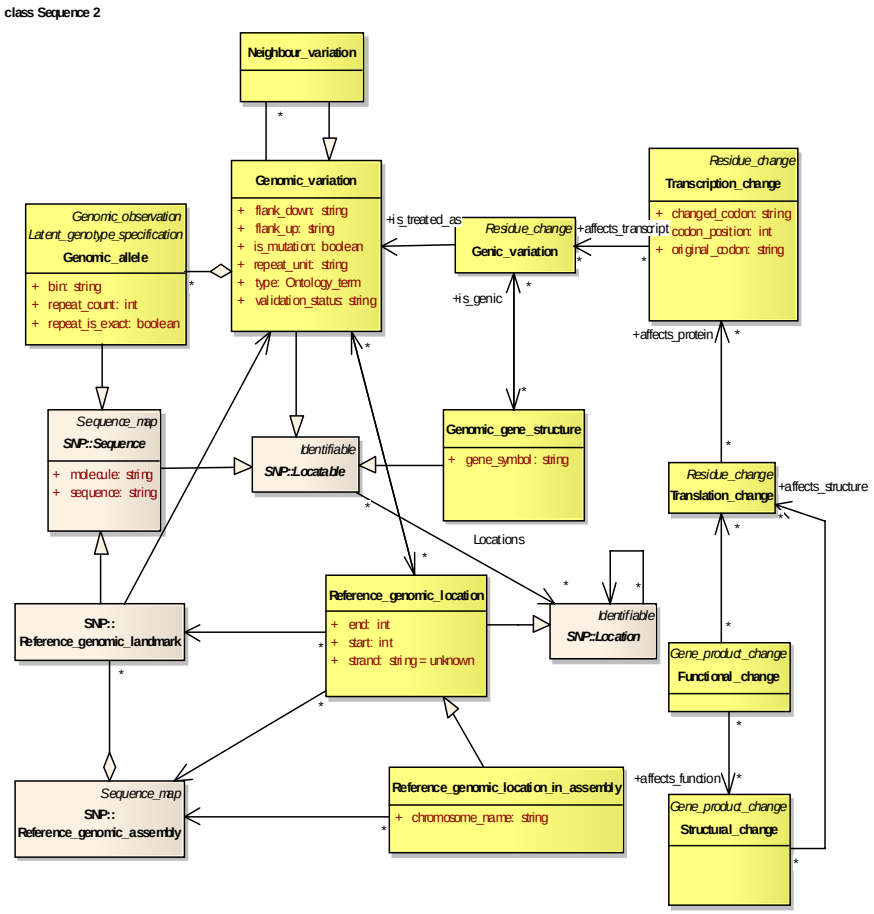

Figure 7.13

Issue 12999 - Attribute “is treated as” renamed to “is_treated_as”

7.1.3.1 Abstract_observable_feature

Type: **Class** **Identifiable**

Package: SNP2

Document:

Abstract extension point for observable (Phenotype) features. Measurable feature of observable (e.g., size of nose).

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Observable_feature	SNP2::Abstract_observable_feature
Aggregation Source -> Destination	SNP2::Abstract_observable_feature	SNP2::Abstract_observation_target
Generalization Source -> Destination	SNP2::Abstract_observable_feature	Identifiable

7.1.3.2 Abstract_observation_target

Type: Class_Identifiable

Package: SNP2

Document:

Abstract_observation_target is abstract super class of observable properties of objects like samples and individuals. Capable of being observed. For example nose size or eye color (Phenotype features).

Connections

Connector	Source	Target
Generalization Source -> Destination	SNP2::Molecular_sample	SNP2::Abstract_observation_target
Association Source -> Destination	SNP2::Assayed_genomic_genotype	SNP2::Abstract_observation_target
Aggregation Source -> Destination	SNP2::Abstract_observable_feature	SNP2::Abstract_observation_target
Association Source -> Destination	PAGE::Location_on_plate	SNP2::Abstract_observation_target
Generalization Source -> Destination	SNP2::Abstract_population	SNP2::Abstract_observation_target
Association Source -> Destination	SNP2::Frequency	SNP2::Abstract_observation_target
Association has value Source -> Destination	PAGE::Observed_value	target SNP2::Abstract_observation_target

Association Source -> Destination	PAGE::Experiment_result	SNP2::Abstract_observation_target
Association Observation targets used in experiment Source -> Destination	PAGE::Genotype_phenotype_correlation_ experiment	SNP2::Abstract_observation_target
Generalization Source -> Destination	SNP2::Abstract_observation_target	Identifiable

7.1.3.3 Abstract_population

Type: Class_Abstract_observation_target

Package: SNP2

Document:

An interbreeding set of individuals, from whom a Panel is drawn (Population in SNP-PML). Extends Abstract_observation_target, which is abstract class for all entities from which one can make genotype or phenotype measurements or observations.

Connections

Connector	Source	Target
Generalization Source -> Destination	SNP2::Abstract_population	SNP2::Abstract_observation_target
Association Source -> Destination	SNP2::Molecular_sample	SNP2::Abstract_population
Association Source -> Destination	SNP2::Abstract_population	Geographic_location
Association Source -> Destination	SNP2::Abstract_population	Taxon
Generalization Source -> Destination	SNP2::Individual	SNP2::Abstract_population
Generalization Source -> Destination	SNP2::Panel	SNP2::Abstract_population

Attributes

Attribute	Notes
ethnicity string	Additional ethnic category of the population sample or “mixed.”
language_family string	Language family name or code, e.g., as in Ethnologue.
primary_language string	Language spoken (name or code), e.g., as in Ethnologue.
race string	Broad ethnic category of the population sample or “mixed.”

7.1.3.4 Abstract_run

Type: Class_Identifiable

Package: SNP2

Document:

Abstract extension point for experiment runs.

Connections

Connector	Source	Target
Generalization Source -> Destination	SNP2::Abstract_run	Identifiable
Association Source -> Destination	SNP2::Assayed_genomic_genotype	SNP2::Abstract_run
Generalization Source -> Destination	PAGE::Run	SNP2::Abstract_run

7.1.3.5 Abstract_value

Type: Class_Identifiable

Package: SNP2

Document:

Abstract class, extension point for values.

Connections

Connector	Source	Target
Association Source -> Destination	PAGE::Defining_feature	SNP2::Abstract_value

Generalization Source -> Destination	PAGE::Value	SNP2::Abstract_value
Generalization Source -> Destination	SNP2::Abstract_value	Identifiable
Dependency derived from raw data Source -> Destination	SNP2::Latent_genotype	SNP2::Abstract_value
Association Source -> Destination	SNP2::Assayed_genomic_genotype	SNP2::Abstract_value
Association Source -> Destination	PAGE::Melting_temperature	SNP2::Abstract_value
Association Source -> Destination	PAGE::Band_size	SNP2::Abstract_value

7.1.3.6 Assayed_genomic_genotype

Type: Class_Genomic_observation

Package: SNP2

Document:

The result of applying a Variation_assay to an individual, to reveal one or more of the Genomic_alleles carried by that individual. This term applies to the observed data rather than to the inferred state of the individual (see Consensus_genomic_genotype). Thus the same individual might have several different Assayed_genomic_genotypes at the same Genomic_variation, where the variation might be due to differing assays, experimental error, dominant systems, missing data, and so forth. Synonym: Measurement.

Association to Genomic_allele is replaced by Latent_genotype, which has possible alleles (Genomic_alleles) as an extension of abstract class Latent_genotype_specification.

Associations from Assayed_genomic_genotype to Value and Run. Value contains raw data like intensity values and Run gives information about experiment run like date and instrument type.

Association from Assayed_genomic_genotype to Molecular_sample is replaced by association to Abstract_observation_target, which is abstract super class of Molecular_sample Assayed_genomic_genotype is now sub-class of Genomic_observation. Genomic_observation is also super class of Frequency and Genomic_Haplotype, and Haplotype_block.

Issue 12999 - Attribute “derived from” renamed to “derived_from”

Connections

Connector	Source	Target
Association Source -> Destination	SNP2::Assayed_genomic_genotype	SNP2::Abstract_observation_target
Association Source -> Destination	SNP2::Assayed_genomic_genotype	SNP2::Abstract_run
Association measured genotype Source -> Destination	SNP2::Assayed_genomic_genotype	SNP2::Latent_genotype
Association without assay details Source -> Destination	SNP2::Assayed_genomic_genotype	SNP2::Genomic_variation
Association with assay details Source -> Destination	SNP2::Assayed_genomic_genotype	SNP2::Variation_assay
Association Source -> Destination	SNP2::Consensus_genomic_genotype	derived_from SNP2::Assayed_genomic_genotype
Association Source -> Destination	PAGE-OM::Page	SNP2::Assayed_genomic_genotype
Generalization Source -> Destination	SNP2::Consensus_genomic_genotype	SNP2::Assayed_genomic_genotype
Association Source -> Destination	SNP2::Assayed_genomic_genotype	SNP2::Abstract_value
Generalization Source -> Destination	SNP2::Assayed_genomic_genotype	SNP2::Genomic_observation

Issue 12999 - Attribute “derived from” renamed to “derived_from”

Attributes

Attribute	Notes
assay_failure boolean	Failure of assay. Value is true if assay has failed.
quality_score float	Quality score of measurement. Depends on the instrument.

7.1.3.7 Consensus_genomic_genotype

Type: Class_Assayed_genomic_genotype

Package: SNP2

Document:

A list of one or more Genomic_alleles carried by a single individual for a single Genomic_variation. Unlike the Assayed_genomic_genotype, the Consensus_genomic_genotype is intended to be used solely for assertions as to the true value of the alleles, after experimental error and other factors are taken into account. The number of Genomic_alleles is equal to the ploidy (copy number) of the individual's genomic DNA at the Reference_genomic_location of the polymorphism. Synonym: Inferred_genomic_genotype, Genomic_genotype.

Issue 12999 - Attribute “derived from” renamed to “derived_from”
13001 - Aggregation from Consensus_genomic_genotype to Latent_genotype is deleted

Connections

Connector	Source	Target
Association Destination -> Source	SNP2::Haplotype_derivation_methods	SNP2::Consensus_genomic_genotype
Association Unspecified	SNP2::Genomic_genotype_population_frequency	SNP2::Consensus_genomic_genotype
Association Source -> Destination	SNP2::Consensus_genomic_genotype	derived_from SNP2::Assayed_genomic_genotype
Generalization Source -> Destination	SNP2::Consensus_genomic_genotype	SNP2::Assayed_genomic_genotype

7.1.3.8 Frequency

Type: Class_Genomic_observation

Package: SNP2

Document:

Abstract class for frequencies, expressed in percentages. Alleles (Genomic_alleles), genotypes (Consensus_genomic_genotype) and haplotypes (Genomic_haplotype) can have measured frequencies in population samples (Panels). In addition, heterozygosity (Heterozygosity) is a measure of observed variability of a polymorphic site (Genomic_variation)in a sub-population (Panel).

Issue 13007 - Association target changed from Panel to Abstract_observation_target

Connections

Connector	Source	Target
Generalization	SNP2::Heterozygosity	SNP2::Frequency

Association Source -> Destination	SNP2::Frequency	SNP2::Abstract_observation_target
Generalization Source -> Destination	SNP2::Frequency	SNP2::Genomic_observation
Generalization Source -> Destination	SNP2::Genomic_haplotype_population_frequency	SNP2::Frequency
Association Source -> Destination	SNP2::Frequency	SNP2::Variation_assay
Generalization Source -> Destination	PAGE::Frequency_set	SNP2::Frequency
Association Source -> Destination	SNP2::Frequency	SNP2::Genomic_variation
Aggregation Source -> Destination	SNP2::Frequency	PAGE::Frequency_set
Generalization Source -> Destination	SNP2::Genomic_allele_population_frequency	SNP2::Frequency
Generalization Source -> Destination	SNP2::Genomic_genotype_population_frequency	SNP2::Frequency

Attributes

Attribute	Notes
count int	Total number
value double	Value of frequency (%)

7.1.3.9 Functional_change

Type: Class_Gene_product_change

Package: SNP2

Document:

Change in the function of the final gene product.

Connections

Connector	Source	Target
Association Source -> Destination	SNP2::Functional_change	SNP2::Translation_change
Association Source -> Destination	SNP2::Functional_change	affects_function SNP2::Structural_change
Association Source -> Destination	PAGE-OM::Page	SNP2::Functional_change
Generalization Source -> Destination	SNP2::Functional_change	Gene_product_change

7.1.3.10 Gene_based_haplotype

Type: Class_Set_of_haplotypes

Package: SNP2

Document:

Collection of variable nucleotides (Genomic_alleles in Genomic_variations) that define a gene. In older usage same as locus.

Connections

Connector	Source	Target
Association Source -> Destination	PAGE-OM::Page	SNP2::Gene_based_haplotype
Generalization Source -> Destination	SNP2::Gene_based_haplotype	SNP2::Set_of_haplotypes

7.1.3.11 Genic_variation

Type: Class_Residue_change

Package: SNP2

Document:

Genomic variation with location in genic coordinates. Synonym: mutation (when change from a common allele affects phenotype).

Issue 12999 - Attribute “is treated as” renamed to “is_treated_as”

Connections

Connector	Source	Target
Generalization	SNP2::Genic_variation	Residue_change
Association Source -> Destination	SNP2::Genomic_gene_structure	SNP2::Genic_variation
Association Source -> Destination	SNP2::Genic_variation	is_treated_as SNP2::Genomic_variation
Association Source -> Destination	SNP2::Transcription_change	affects_transcript SNP2::Genic_variation
Association Source -> Destination	is_genic SNP2::Genic_variation	SNP2::Genomic_gene_structure
Association Source -> Destination	PAGE-OM::Page	SNP2::Genic_variation

7.1.3.12 Genomic_allele

Issue 13010 - New inheritance from Genomic_observable

Type: Class_Genomic_observation, Latent_genotype_specification, Sequence

Package: SNP2

Document:

One of several alternative DNA sequences of a Reference_genomic_location as it appears in the population of organisms. Synonym: variant, allele

Issue 12999 - Attribute “one of” renamed to “one_of”

Connections

Connector	Source	Target
Generalization	Genomic_reference_allele	SNP2::Genomic_allele
Association Unspecified	SNP2::Genomic_allele	SNP2::Genomic_haplotype

Aggregation Source -> Destination	SNP2::Genomic_allele	SNP2::Genomic_variation
Generalization Source -> Destination	SNP2::Genomic_allele	SNP2::Latent_genotype_specification
Generalization	SNP2::Genomic_allele	Sequence
Generalization Source -> Destination	SNP2::Genomic_allele	SNP2::Genomic_observation
Association Source -> Destination	one_of Genomic_reference_allele	SNP2::Genomic_allele
Association Source -> Destination	PAGE-OM::Page	SNP2::Genomic_allele
Association Source -> Destination	SNP2::Genomic_allele_population_frequency	SNP2::Genomic_allele

Attributes

Attribute	Notes
bin string	Size class for microsatellite alleles when exact size cannot be determined. Semantic name.
repeat_count int	If the Genomic_variation type is 'microsatellite,' get number of repeat units as value, e.g., 7.
repeat_is_exact boolean	The Genomic_variation type is 'microsatellite' and gets value true if the allele region consists of repeat units only.

7.1.3.13 Genomic_allele_population_frequency

Type: Class_Frequency

Package: SNP2

Document:

The frequency with which a particular Genomic_allele is seen in a particular Panel. This frequency can be measured from pooled samples. Synonyms: Genomic_allele_panel_frequency, allele_frequency.

Connections

Connector	Source	Target
Association Source -> Destination	PAGE-OM::Page	SNP2::Genomic_allele_population_ frequency
Association Source -> Destination	SNP2::Genomic_allele_population_frequency	SNP2::Genomic_allele
Generalization Source -> Destination	SNP2::Genomic_allele_population_frequency	SNP2::Frequency

7.1.3.14 Genomic_gene_structure

Type: Class_Locatable

Package: SNP2

Document:

A structure of a gene expressed as location of the CDS and exons. Defines genic coordinate system from start of the CDS downstream.

Connections

Connector	Source	Target
Association Unspecified	Exon	SNP2::Genomic_gene_structure
Association Unspecified	CDS	SNP2::Genomic_gene_structure
Generalization	SNP2::Genomic_gene_structure	Locatable
Association Source -> Destination	SNP2::Genomic_gene_structure	SNP2::Genic_variation
Association Source -> Destination	is_genic SNP2::Genic_variation	SNP2::Genomic_gene_structure
Association Source -> Destination	PAGE-OM::Page	SNP2::Genomic_gene_structure

Attributes

Attribute	Notes
gene_symbol string	Gene symbol for the gene (e.g., approved by the HUGO nomenclature committee).

7.1.3.15 Genomic_genotype_population_frequency

Type: Class_Frequency

Package: SNP2

Document:

Frequency of a Consensus_genomic_genotype in a Panel.

OSAGE-OM

Has many to one relationship to Latent_genotype (Consensus_genomic_genotype in SNP-PML).

Connections

Connector	Source	Target
Association measured genotype Source -> Destination	SNP2::Genomic_genotype_population_frequency	SNP2::Latent_genotype
Association Unspecified	SNP2::Genomic_genotype_population_frequency	SNP2::Consensus_genomic_genotype
Association Source -> Destination	PAGE-OM::Page	SNP2::Genomic_genotype_population_frequency
Generalization Source -> Destination	SNP2::Genomic_genotype_population_frequency	SNP2::Frequency

7.1.3.16 Genomic_haplotype

Type: Class_Genomic_observation

Package: SNP2

Document:

A set of Genomic_alleles across an equal number of Genomic_variations in a single chromosome and in a single individual. The Genomic_haplotype is derived from a set of Consensus_genomic_genotype. For each Genomic_variation, the haplotype contains one and only one Genomic_allele. Furthermore, the Genomic_alleles are required to be in phase on the individual, meaning that they are located on the same contiguous strand of DNA. Synonym: Haplotype.

Connections

Connector	Source	Target
Association Unspecified	SNP2::Genomic_allele	SNP2::Genomic_haplotype
Association Destination -> Source	SNP2::Genomic_haplotype	SNP2::Haplotype_derivation_methods
Association Unspecified	SNP2::Genomic_haplotype	SNP2::Set_of_haplotypes
Generalization Source -> Destination	SNP2::Genomic_haplotype	SNP2::Genomic_observation
Association Source -> Destination	SNP2::Genomic_haplotype	SNP2::Molecular_sample
Association Source -> Destination	SNP2::Haplotype_map	SNP2::Genomic_haplotype
Association Source -> Destination	PAGE-OM::Page	SNP2::Genomic_haplotype
Association Source -> Destination	SNP2::Genomic_haplotype_population_frequency	SNP2::Genomic_haplotype
Association Source -> Destination	SNP2::Genomic_haplotype	SNP2::Haplotype_map

7.1.3.17 Genomic_haplotype_population_frequency

Type: Class_Frequency

Package: SNP2

Document:

Frequency of a Genomic_haplotype in a Panel.

Connections

Connector	Source	Target
Association Source -> Destination	PAGE-OM::Page	SNP2::Genomic_haplotype_population_frequency

Generalization Source -> Destination	SNP2::Genomic_haplotype_population_frequency	SNP2::Frequency
Association Source -> Destination	SNP2::Genomic_haplotype_population_frequency	SNP2::Genomic_haplotype

7.1.3.18 Genomic_observation

Type: Class_Identifiable

Package: SNP2

Document:

Genomic observation

Connections

Issue 13010 - New generalization from Genomic_allele to Genomic_observation

Connector	Source	Target
Generalization Source -> Destination	SNP2::Genomic_haplotype	SNP2::Genomic_observation
Generalization Source -> Destination	SNP2::Genomic_allele	SNP2::Genomic_observation
Generalization Source -> Destination	SNP2::Frequency	SNP2::Genomic_observation
Association Source -> Destination	PAGE::Experiment_result	SNP2::Genomic_observation
Association Source -> Destination	PAGE::Genotype_phenotype_correlation_experiment	SNP2::Genomic_observation
Generalization Source -> Destination	SNP2::Genomic_observation	Identifiable
Generalization Source -> Destination	SNP2::Set_of_haplotypes	SNP2::Genomic_observation

Generalization Source -> Destination	SNP2::Assayed_genomic_genotype	SNP2::Genomic_observation

7.1.3.19 Genomic_variation

Type: Class_Locatable

Package: SNP2

Document:

A variable site in a Reference_genomic_landmark sequence. Synonyms: polymorphic site, marker, (Genomic_polymorphism in SNP-PML). The relationship to Latent_genotype_specification gives information on possible sequence variations attached to the locus (as defined by flanking sequences).

Issue 12999 - Attribute “is treated as” renamed to “is_treated_as”
13003 - Association Multi-variation_assay to Genomic_variation is remove

Connections

Connector	Source	Target
Generalization	SNP2::Neighbour_variation	SNP2::Genomic_variation
Aggregation Source -> Destination	SNP2::Genomic_allele	SNP2::Genomic_variation
Association Source -> Destination	SNP2::Reference_genomic_location	SNP2::Genomic_variation
Association Destination -> Source	SNP2::Genomic_variation	Reference_genomic_landmark
Association Source -> Destination	SNP2::Variation_assay	SNP2::Genomic_variation
Association Source -> Destination	SNP2::Genomic_variation	SNP2::Reference_genomic_location
Association Source -> Destination	SNP2::Genic_variation	is_treated_as SNP2::Genomic_variation
Association Destination -> Source	Ordered_location	SNP2::Genomic_variation
Association without assay details Source -> Destination	SNP2::Assayed_genomic_genotype	SNP2::Genomic_variation

Association Unspecified	SNP2::Genomic_variation	SNP2::Heterozygosity
Association Unspecified	SNP2::Genomic_variation	SNP2::Neighbour_variation
Generalization Source -> Destination	SNP2::Genomic_variation	Locatable
Association Source -> Destination	PAGE-OM::Page	SNP2::Genomic_variation
Association Source -> Destination	SNP2::Frequency	SNP2::Genomic_variation
Association Genomic variations used in experiment Source -> Destination	PAGE::Genotype_phenotype_correlation_experiment	SNP2::Genomic_variation
Association Source -> Destination	PAGE::Experiment_result	SNP2::Genomic_variation

Attributes

Attribute	Notes
flank_down string	Downstream flanking sequence (at least 25 residues, if possible).
flank_up string	Upstream flanking sequence (at least 25 residues, if possible).
is_mutation boolean	Proven phenotype change causing mutation.
repeat_unit string	If type is 'microsatellite', gives the repeat unit, e.g., “CA.”
type Ontology_term	The type of the polymorphism (e.g., SNP, microsatellite, indel, translocation,...).
validation_status string	Validation status, e.g., “Proven,” “Suspected.”

7.1.3.20 Haplotype_block

Type: Class_Set_of_haplotypes

Package: SNP2

Document:

Large (spanning a few kb to >100 kb) blocks of Genomic_alleles in linkage disequilibrium (LD) and a few haplotypes per block, separated by regions of recombination.

Connections

Connector	Source	Target
Generalization	SNP2::Haplotype_block	SNP2::Set_of_haplotypes
Association Source -> Destination	PAGE-OM::Page	SNP2::Haplotype_block

7.1.3.21 Haplotype_derivation_methods

Type: Class_Methodology

Package: SNP2

Document:

Association class describing methods used to derive Genomic_haplotypes from Consensus_genomic_genotypes.

Connections

Connector	Source	Target
Association Destination -> Source	SNP2::Genomic_haplotype	SNP2::Haplotype_derivation_methods
Association Destination -> Source	SNP2::Haplotype_derivation_methods	SNP2::Consensus_genomic_genotype
Association Source -> Destination	PAGE-OM::Page	SNP2::Haplotype_derivation_methods
Generalization Source -> Destination	SNP2::Haplotype_derivation_methods	Methodology

7.1.3.22 Haplotype_map

Type: Class_Physical_map

Package: SNP2

Document:

Map of haplotypes. Features include: * Block length distribution * measures of block variability * relative proportions of common haplotypes * block coverage of chromosomes and/or genome * LD and other values between haplotypes, markers, alleles

Connections

Connector	Source	Target
Generalization	SNP2::Haplotype_map	Physical_map
Association Unspecified	SNP2::Haplotype_map	SNP2::Set_of_haplotypes
Association Source -> Destination	SNP2::Haplotype_map	SNP2::Genomic_haplotype
Association Source -> Destination	PAGE-OM::Page	SNP2::Haplotype_map
Association Source -> Destination	SNP2::Genomic_haplotype	SNP2::Haplotype_map

7.1.3.23 Heterozygosity

Type: Class_Frequency

Package: SNP2

Document:

A numerical measure of observed abundance of different Genomic_alleles in a Genomic_variation.

Connections

Connector	Source	Target
Generalization	SNP2::Heterozygosity	SNP2::Frequency
Association Unspecified	SNP2::Genomic_variation	SNP2::Heterozygosity
Association Source -> Destination	PAGE-OM::Page	SNP2::Heterozygosity

7.1.3.24 Individual

Type: Class_Abstract_population

Package: SNP2

Document:

A single member of a species, where a species is an accessioned taxon defined by a public database, and the individual is accessioned in a public or private database. Synonym: “inbred strain” in homozygous lineages.

Connections

Connector	Source	Target
Association Unspecified	SNP2::Individual	SNP2::Panel
Generalization Source -> Destination	SNP2::Individual	SNP2::Abstract_population
Association Source -> Destination	PAGE-OM::Page	SNP2::Individual

Issue 13008 - Attributed sex changed to gender

Attributes

Attribute	Notes
birth_date string	Date of birth of the individual. May be better abbreviated to birth year to protect the privacy of the individual
death_date string	Date of death of the individual. May be better abbreviated to plain year to protect the privacy of the individual.
father_id string	Id of the father to allow building of pedigrees.
gender string	Recommended values are ‘unknown,’ ‘male,’ and ‘female.’ Additional values can be used to denote unusual karyotypes.
mother_id string	Id of the mother to allow building of pedigrees.

7.1.3.25 Latent_genotype

Type: Class_Identifiable

Package: SNP2

Document:

Potentially existing genotypes that could be observed by Variation_assays (Polymorphism_assays in SNP-PML) Application of a Variation_assay on one Molecular_sample generates a single Latent_genotype which has one or more Latent_genotype_specifications (this depends on ploidy level in case of Genomic_allele).

Latent_genotypes associated to one instance of a Variation_assay can have only one type of Latent_genotype_specifications, as defined by Defining_feature.

Class is a holder for one or many observable variation objects (Latent_genotype_specification). Latent_genotype is used to attach possible variations to measurements (Assayed_genomic_genotype), variation assays (Variation_assay) and marker loci (Genomic_variation). The construct is used in places where Genomic_allele was used in the SNP-PML.

Issue 13001 - Aggregation from Consensus_geomic_genotype to Latent_genotype is deleted

Connections

Connector	Source	Target
Generalization Source -> Destination	SNP2::Latent_genotype	Identifiable
AssociationClass Unspecified	SNP2::Latent_genotype	SNP2::Latent_genotype_specification
Association detectable genotypes Source -> Destination	SNP2::Variation_assay	SNP2::Latent_genotype
Association measured genotype Source -> Destination	SNP2::Assayed_genomic_genotype	SNP2::Latent_genotype
Association measured genotype Source -> Destination	SNP2::Genomic_genotype_population_frequency	SNP2::Latent_genotype
Association Source -> Destination	PAGE-OM::Page	SNP2::Latent_genotype
Dependency derived from raw data Source -> Destination	SNP2::Latent_genotype	SNP2::Abstract_value

7.1.3.26 Latent_genotype_specification

Type: Class

Package: SNP2

Document:

Abstract super class of observable variation objects, like alleles, melting temperatures (Melting_temperature), band sizes (Band_size). The class makes possible to extend the model to cover different kind of sequence or chromosomal variations.

Connections

Connector	Source	Target
AssociationClass Unspecified	SNP2::Latent_genotype	SNP2::Latent_genotype_specification

Generalization Source -> Destination	PAGE::Melting_temperature	SNP2::Latent_genotype_specification
Generalization Source -> Destination	PAGE::Band_size	SNP2::Latent_genotype_specification
Generalization Source -> Destination	SNP2::Genomic_allele	SNP2::Latent_genotype_specification
Association Source -> Destination	PAGE::Defining_feature	SNP2::Latent_genotype_specification

7.1.3.27 Molecular_sample

Type: Class_Abstract_observation_target

Package: SNP2

Document:

A sample from an Individual or from a Panel defining the molecule and tissue/cell used (Anatomic_locations) in the Variation_assay. Synonym: Sample_of_individual.

Class extends the Abstract_observation_target, which is abstract class for all entities from which one can make genotype or phenotype measurements or observations.

Connections

Connector	Source	Target
Association Source -> Destination	SNP2::Molecular_sample	SNP2::Molecular_sample
Generalization Source -> Destination	SNP2::Molecular_sample	SNP2::Abstract_observation_target
Association Source -> Destination	SNP2::Molecular_sample	SNP2::Abstract_population
Association Source -> Destination	SNP2::Genomic_haplotype	SNP2::Molecular_sample
Association Source -> Destination	SNP2::Molecular_sample	Anatomic_location
Association Source -> Destination	PAGE-OM::Page	SNP2::Molecular_sample

Attributes

Attribute	Notes
molecule string	The molecule (RNA, DNA, protein) used in the assay.

7.1.3.28 Neighbour_variation

Type: Class_Genomic_variation

Package: SNP2

Document:

Another Genomic_variation close enough to affect the primer design.

Connections

Connector	Source	Target
Generalization	SNP2::Neighbour_variation	SNP2::Genomic_variation
Association Unspecified	SNP2::Genomic_variation	SNP2::Neighbour_variation
Association Source -> Destination	PAGE-OM::Page	SNP2::Neighbour_variation

7.1.3.29 Panel

Type: Class_Abstract_population

Package: SNP2

Document:

A set of samples from individuals drawn from the same species and used for genetic studies. A panel must be identifiable with a list of accessioned individuals, if possible. (Note: do not use term “population” here!) Synonym: SampleSet, Sample from population(s), “Plate” in Coriel sense. Recursion. Panels of panels.

Connections

Connector	Source	Target
Association Unspecified	SNP2::Individual	SNP2::Panel
Association Source -> Destination	SNP2::Panel	SNP2::Panel

Association Source -> Destination	PAGE-OM::Page	SNP2::Panel
Generalization Source -> Destination	SNP2::Panel	SNP2::Abstract_population

Attributes

Attribute	Notes
count_unit string individual	Values are ‘chromosome’ and ‘individual.’ Default is ‘individual.’
pooled boolean	True if accessioned individuals are not available.
size long	The size of the sample. Note that the count_unit field affects how this value is interpreted.
type string	Optional identifier of the panel category: e.g., plate, family, population sample.

7.1.3.30 Reference_genomic_location

Type: Class_Location

Package: SNP2

Document:

A location within a Reference_genomic_landmark. Attributes of the location are the Reference_genomic_assembly and/or the Reference_genomic_landmark, the start and end range and strand of the feature relative to the Reference_genomic_landmark.

Connections

Connector	Source	Target
Association Destination -> Source	Reference_genomic_assembly	SNP2::Reference_genomic_location
Association Destination -> Source	Reference_genomic_landmark	SNP2::Reference_genomic_location
Generalization	SNP2::Reference_genomic_location_in_assembly	SNP2::Reference_genomic_location
Generalization	SNP2::Reference_genomic_location	Location
Association Source -> Destination	SNP2::Reference_genomic_location	SNP2::Genomic_variation

Association Source -> Destination	SNP2::Genomic_variation	SNP2::Reference_genomic_location
Association Source -> Destination	PAGE-OM::Page	SNP2::Reference_genomic_location

Attributes

Attribute	Notes
end int	End of the location in the reference sequence.
start int	Start of the location in the reference sequence.
strand string unknown	Orientation of the feature in the reference sequence. One of 'forward,''reverse,' 'unknown.' Defaults to 'unknown.'

7.1.3.31 Reference_genomic_location_in_assembly

Type: Class_Reference_genomic_location

Package: SNP2

Document:

A location in one chromosome of a reference genomic assembly. Instead of the reference sequence being an accessioned sequence, it is a versioned assembly.

Connections

Connector	Source	Target
Generalization	SNP2::Reference_genomic_location_in_assembly	SNP2::Reference_genomic_location
Association Destination -> Source	Reference_genomic_assembly	SNP2::Reference_genomic_location_in_assembly

Attributes

Attribute	Notes
chromosome_name string	Name of the chromosome in the assembly.

7.1.3.32 Set_of_haplotypes

Issue 13002 - Redundant inheritance from Identifiable is removed in Set_of_haplotypes

Type: Class_Genomic_observation

Package: SNP2

Document:

An abstract representation of a collection of haplotypes.

Connections

Connector	Source	Target
Generalization	SNP2::Haplotype_block	SNP2::Set_of_haplotypes
Association Unspecified	SNP2::Genomic_haplotype	SNP2::Set_of_haplotypes
Association Unspecified	SNP2::Haplotype_map	SNP2::Set_of_haplotypes
Generalization Source -> Destination	SNP2::Gene_based_haplotype	SNP2::Set_of_haplotypes
Generalization Source -> Destination	SNP2::Set_of_haplotypes	SNP2::Genomic_observation

7.1.3.33 Structural_change

Type: Class_Gene_product_change

Package: SNP2

Document:

Change in the 3D structure of the polypeptide chain.

Connections

Connector	Source	Target
Generalization Source -> Destination	SNP2::Structural_change	Gene_product_change
Association Source -> Destination	SNP2::Functional_change	affects_function SNP2::Structural_change

Association Source -> Destination	SNP2::Structural_change	affects_structure SNP2::Translation_change
Association Source -> Destination	PAGE-OM::Page	SNP2::Structural_change

7.1.3.34 Transcription_change

Type: Class_Residue_change

Package: SNP2

Document:

Change in the quality or quantity of the mature RNA product.

Connections

Connector	Source	Target
Association Source -> Destination	SNP2::Translation_change	affects_protein SNP2::Transcription_change
Association Source -> Destination	SNP2::Transcription_change	affects_transcript SNP2::Genic_variation
Association Source -> Destination	PAGE-OM::Page	SNP2::Transcription_change
Generalization Source -> Destination	SNP2::Transcription_change	Residue_change

Attributes

Attribute	Notes
changed_codon string	The new codon in the transcript, if applicable.
codon_position int	The first affected nucleotide in the codon. Values are: 1, 2, or 3.
original_codon string	The affected codon in the transcript.

7.1.3.35 Translation_change

Type: Class_Residue_change

Package: SNP2

Document:

Change in the quality or quantity of (predicted) polypeptide chain (2D).

Connections

Connector	Source	Target
Generalization	SNP2::Translation_change	Residue_change
Association Source -> Destination	SNP2::Functional_change	SNP2::Translation_change
Association Source -> Destination	SNP2::Structural_change	affects_structure SNP2::Translation_change
Association Source -> Destination	SNP2::Translation_change	affects_protein SNP2::Transcription_change
Association Source -> Destination	PAGE-OM::Page	SNP2::Translation_change

7.1.3.36 Variation_assay

Type: Class_Methodology

Package: SNP2

Document:

An experimental lab protocol and set of reagents for detecting the Genomic_alleles of Genomic_variations carried by an individual or a panel of individuals. Synonym: Assay. Non instrument part of the experiment - same assay can be used in different instrument runs. (Polymorphism_assay in SNP-PML).

Variation assay designed for the Genomic_variation has one-to-many association to Latent_genotype_specification, which gives information on measurable variations (by this assay) attached to the locus.

Connections

Connector	Source	Target
Generalization Source -> Destination	PAGE::Multi_vartiation_assay	SNP2::Variation_assay
Generalization	SNP2::Variation_assay	Methodology
Association detectable genotypes Source -> Destination	SNP2::Variation_assay	SNP2::Latent_genotype

Association Source -> Destination	SNP2::Variation_assay	SNP2::Genomic_variation
Aggregation Source -> Destination	SNP2::Variation_assay	PAGE::Multi_vartiation_assay
Association Source -> Destination	SNP2::Variation_assay	Oligo
Association With assay details Source -> Destination	SNP2::Assayed_genomic_genotype	SNP2::Variation_assay
Association Source -> Destination	PAGE-OM::Page	SNP2::Variation_assay
Association Source -> Destination	SNP2::Frequency	SNP2::Variation_assay

Attributes

Attribute	Notes
description string	Free text description of the assay protocol.

7.1.4 FUGE

Type: Package «XSDschema»

Package: PAGE-OM

Ontology - (Logical diagram)

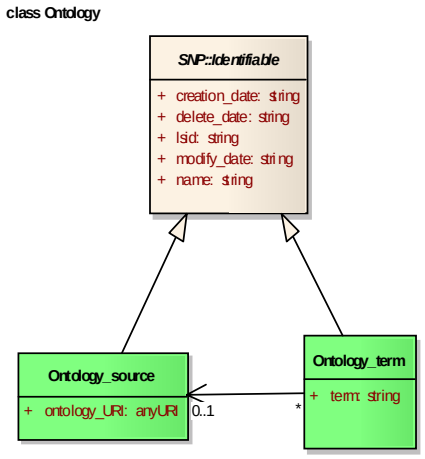

Figure 7.14

7.1.4.1 Ontology_source

Type: Class_Identifiable

Package: FUGE

Document:

The source ontology or controlled vocabulary list that ontology terms have been obtained from.

Connections

Connector	Source	Target
Association Source -> Destination	FUGE::Ontology_term	FUGE::Ontology_source
Generalization Source -> Destination	FUGE::Ontology_source	Identifiable

Attributes

Attribute	Notes
ontology_URI anyURI	A URI is short for Uniform Resource Identifier. A URI is a compact sequence of characters that identifies an abstract or physical resource.

7.1.4.2 Ontology_term

Type: Class_Identifiable

Package: FUGE

Document:

Ontology term, defined in more detail in FuGE (http://fuge.sourceforge.net/). This is just a place holder.

Connections

Connector	Source	Target
Association Source -> Destination	FUGE::Ontology_term	FUGE::Ontology_source
Generalization Source -> Destination	FUGE::Ontology_term	Identifiable

Attributes

Attribute	Notes
term string	Ontology term

7.1.5 BasicTypes

Type: Package «XSDschema»

Package: PAGE-OM

BasicTypes - *(Logical diagram)*

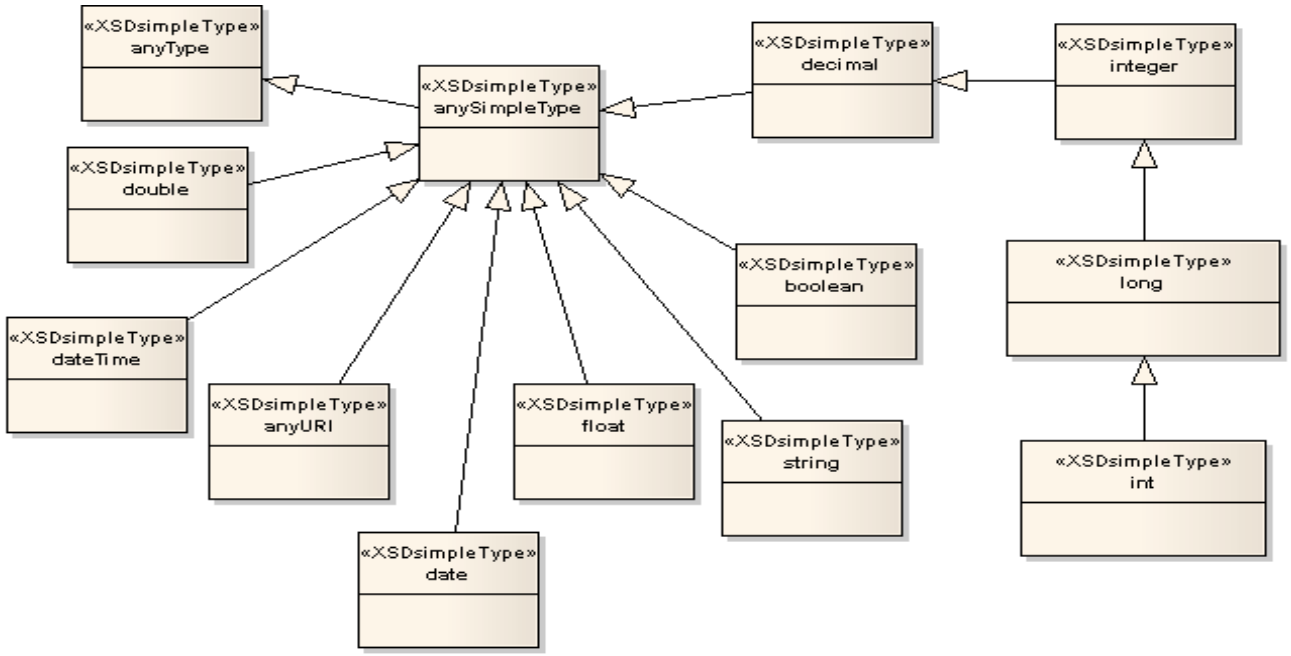

Figure 7.15

7.1.5.1 anySimpleType

Type: Class_anyType

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::decimal	BasicTypes::anySimpleType
Generalization	BasicTypes::float	BasicTypes::anySimpleType
Generalization	BasicTypes::anyURI	BasicTypes::anySimpleType
Generalization	BasicTypes::dateTime	BasicTypes::anySimpleType
Generalization	BasicTypes::date	BasicTypes::anySimpleType

Generalization	BasicTypes::double	BasicTypes::anySimpleType
Generalization	BasicTypes::anySimpleType	BasicTypes::anyType
Generalization	BasicTypes::string	BasicTypes::anySimpleType
Generalization	BasicTypes::time	BasicTypes::anySimpleType
Generalization	BasicTypes::boolean	BasicTypes::anySimpleType

7.1.5.2 anyType

Type: Class

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::anySimpleType	BasicTypes::anyType

7.1.5.3 anyURI

Type: Class_anySimpleType

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::anyURI	BasicTypes::anySimpleType

7.1.5.4 boolean

Type: Class_anySimpleType

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::boolean	BasicTypes::anySimpleType

7.1.5.5 date

Type: Class_anySimpleType

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::date	BasicTypes::anySimpleType

7.1.5.6 dateTime

Type: Class_anySimpleType

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::dateTime	BasicTypes::anySimpleType

7.1.5.7 decimal

Type: Class_anySimpleType

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::decimal	BasicTypes::anySimpleType
Generalization	BasicTypes::integer	BasicTypes::decimal

7.1.5.8 double

Type: Class_anySimpleType

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::double	BasicTypes::anySimpleType

7.1.5.9 float

Type: Class_anySimpleType

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::float	BasicTypes::anySimpleType

7.1.5.10 int

Type: Class_long

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::short	BasicTypes::int
Generalization	BasicTypes::int	BasicTypes::long

7.1.5.11 integer

Type: Class_decimal

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::long	BasicTypes::integer
Generalization	BasicTypes::integer	BasicTypes::decimal

7.1.5.12 long

Type: Class_integer

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::int	BasicTypes::long

Generalization	BasicTypes::long	BasicTypes::integer

7.1.5.13 short

Type: Class_int

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::short	BasicTypes::int

7.1.5.14 string

Type: Class_anySimpleType

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::string	BasicTypes::anySimpleType

7.1.5.15 time

Type: Class_anySimpleType

Package: BasicTypes

Document:

Connections

Connector	Source	Target
Generalization	BasicTypes::time	BasicTypes::anySimpleType

7.1.6 SNP

Type: Package «XSDschema»

Package: PAGE-OM

Map - *(Logical diagram)*

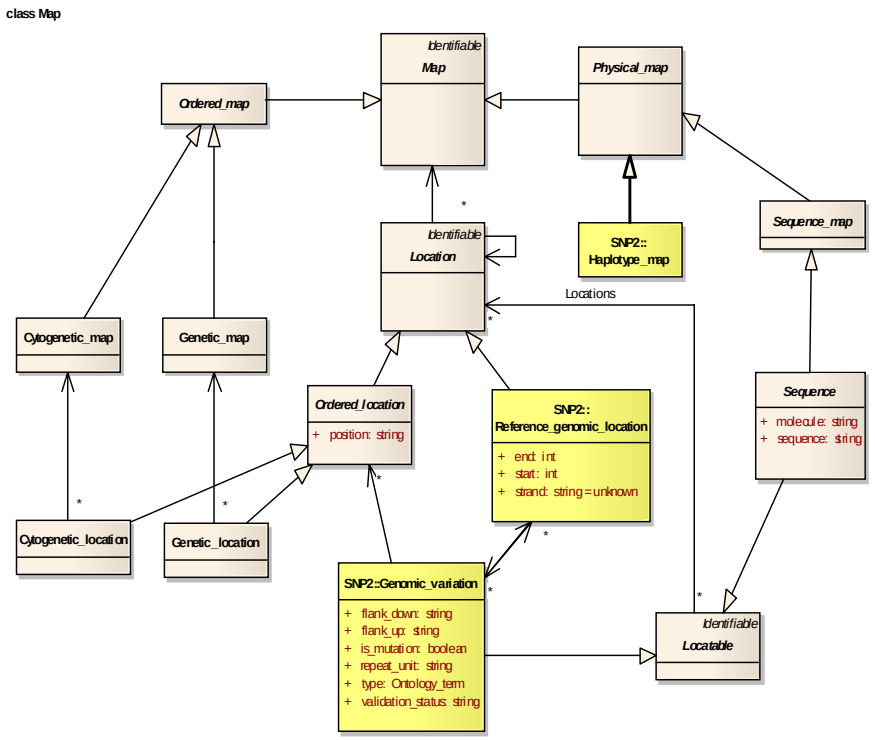

Figure 7.16

Sample - (*Logical diagram*)

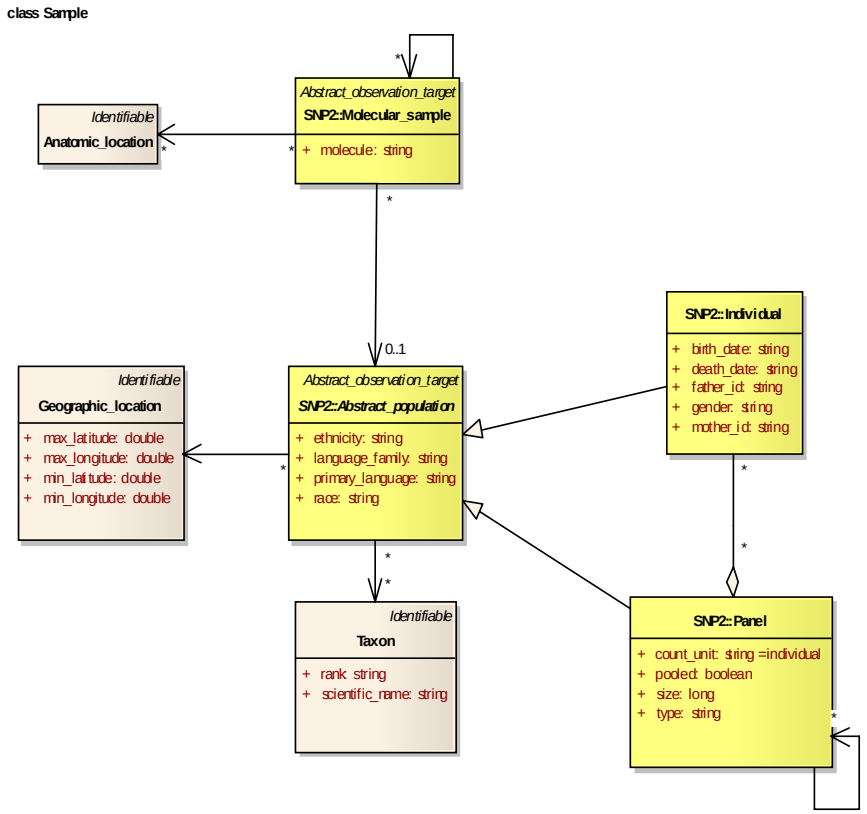

Figure 7.17

Issue 13008 - Attribute sex in Individual is renamed to gender

Sequence - (*Logical diagram*)

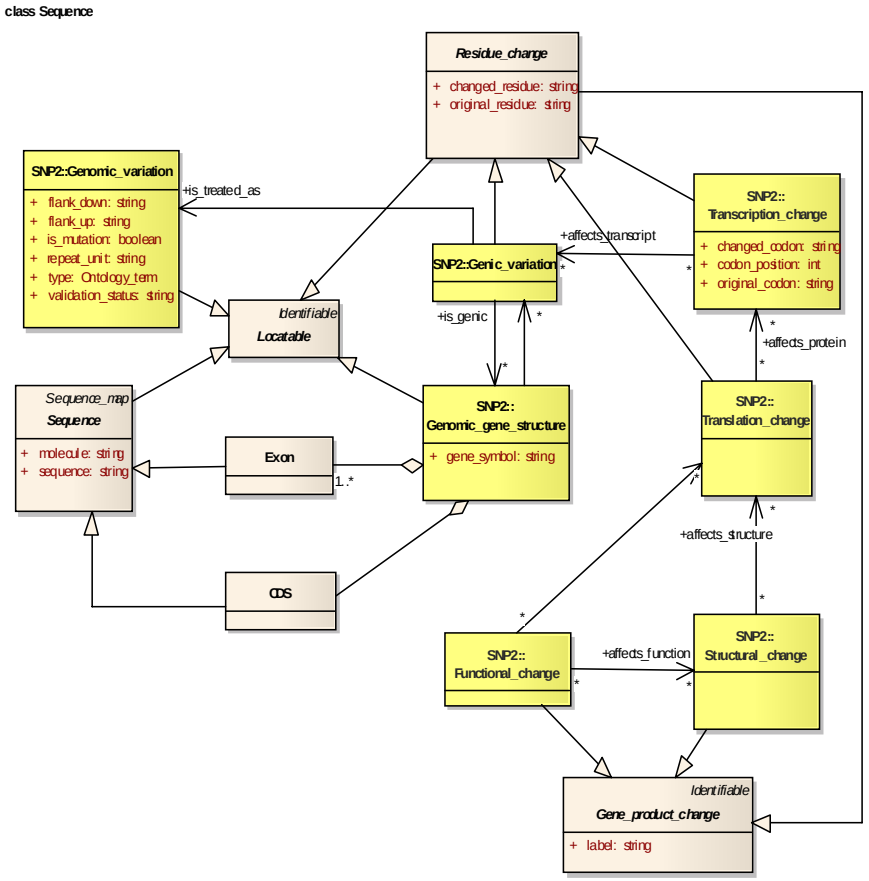

Figure 7.18

Issue 12999 - Attribute “is treated as” renamed to “is_treated_as”

Sequence 2 - (*Logical diagram*)

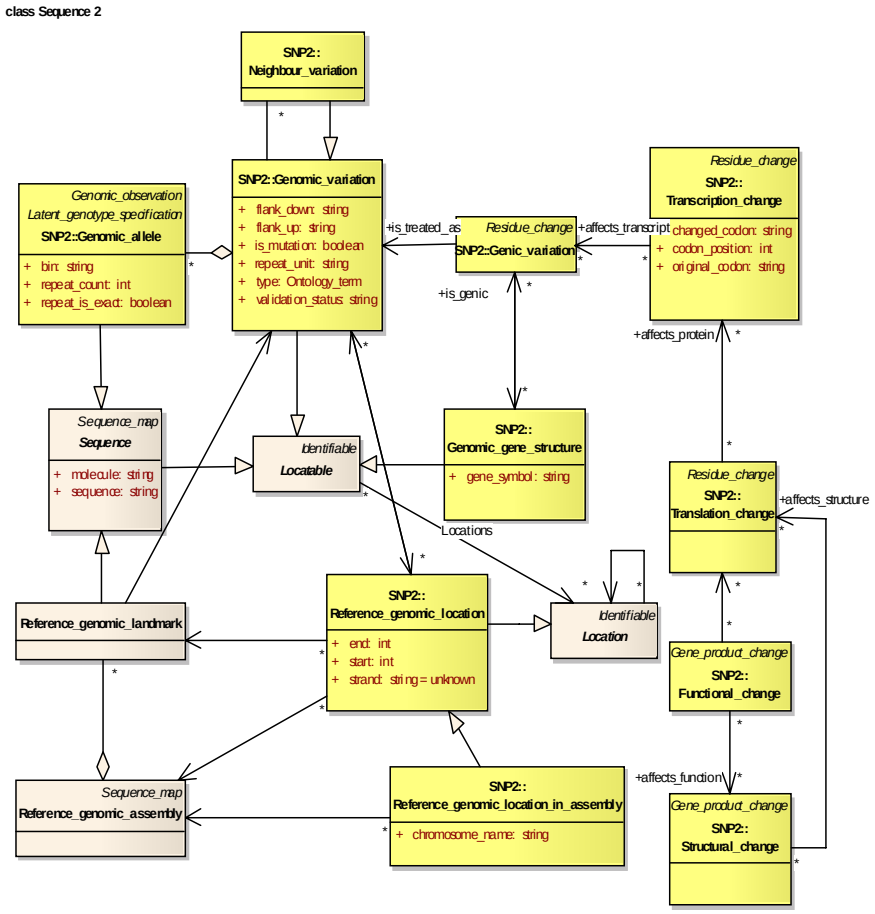

Figure 7.19

Issue 12999 - Attribute “is treated as” renamed to “is_treated_as”

7.1.6.1 Anatomic_location

Type: Class_Identifiable

Package: SNP

Document:

Description of the anatomical location the molecular sample is derived from. Best expressed as a controlled vocabulary of anatomical terms.

Connections

Connector	Source	Target
Association Source -> Destination	SNP2::Molecular_sample	Anatomic_location
Association Source -> Destination	PAGE-OM::Page	Anatomic_location
Generalization Source -> Destination	Anatomic_location	Identifiable

7.1.6.2 Annotation

Type: Class

Package: SNP

Document:

A free text of name and value pair. Note: name should be Ontology_term.

Connections

Connector	Source	Target
Association Source -> Destination	Identifiable	Annotation
Association Source -> Destination	PAGE-OM::Page	Annotation

Attributes

Attribute	Notes
name string	Name of the annotation feature
value Value	Value of annotation feature

7.1.6.3 CDS

Type: Class_Sequence

Package: SNP

Document:

Region in a reference sequence that determines the start, end, and strand of the gene coding region.

Connections

Connector	Source	Target
Association Unspecified	CDS	SNP2::Genomic_gene_structure
Generalization	CDS	Sequence
Association Source -> Destination	PAGE-OM::Page	CDS

7.1.6.4 Contributor

Type: Class_Source

Package: SNP

Document:

Person who has edited data in the data set. Synonym: curator.

Connections

Connector	Source	Target
Association Source -> Destination	PAGE-OM::Page	Contributor
Generalization Source -> Destination	Contributor	BREF::Source

Attributes

Attribute	Notes
date string	Date of the editing.

7.1.6.5 Cytogenetic_location

Type: Class_Ordered_location

Package: SNP

Document:

A location in a cytogenetic map expressed as a band or a range of bands.

Connections

Connector	Source	Target
Association Destination -> Source	Cytogenetic_map	Cytogenetic_location
Generalization	Cytogenetic_location	Ordered_location
Association Source -> Destination	PAGE-OM::Page	Cytogenetic_location

7.1.6.6 Cytogenetic_map

Type: Class_Ordered_map

Package: SNP

Document:

Map of cytogenetic bands describing their relative order.

Connections

Connector	Source	Target
Association Destination -> Source	Cytogenetic_map	Cytogenetic_location
Generalization	Cytogenetic_map	Ordered_map
Association Source -> Destination	PAGE-OM::Page	Cytogenetic_map

7.1.6.7 Db_xref

Type: Class_Identifiable

Package: SNP

Document:

Cross reference between two Identifiables in different databases.

Connections

Connector	Source	Target
Association Source -> Destination	Identifiable	Db_xref
Association Destination -> Source	identifier Db_xref	BREF::Bibliographic_reference
Association Source -> Destination	PAGE-OM::Page	Db_xref
Generalization Source -> Destination	Db_xref	Identifiable

Attributes

Attribute	Notes
db string	The remote database name or abbreviation.
db_version string	Version of the database
field string	Field in the remote database where the UID value is found (e.g., ID or AC in EMBL). Optional
uid string	Unique identifier in the remote database.
url string	Full URL to the cross-referenced entry

7.1.6.8 Exon

Type: Class_Sequence

Package: SNP

Document:

Region in a reference sequence that determines the start, end, and strand of the transcript that is not spliced away.

Connections

Connector	Source	Target
Association Unspecified	Exon	SNP2::Genomic_gene_structure
Association Source -> Destination	PAGE-OM::Page	Exon

Generalization Source -> Destination	Exon	Sequence

7.1.6.9 Gene_product_change

Type: Class_Identifiable

Package: SNP

Document:

Free form description of change in gene product

Connections

Connector	Source	Target
Generalization Source -> Destination	SNP2::Structural_change	Gene_product_change
Generalization Source -> Destination	Gene_product_change	Identifiable
Generalization Source -> Destination	Residue_change	Gene_product_change
Generalization Source -> Destination	SNP2::Functional_change	Gene_product_change

Attributes

Attribute	Notes
label string	Human understandable term for the change (e.g., missense, truncation).

7.1.6.10 Genetic_location

Type: Class_Ordered_location

Package: SNP

Document:

A relative location of a marker in a genetic map based on recombination.

Connections

Connector	Source	Target
Association Destination -> Source	Genetic_map	Genetic_location
Generalization Source -> Destination	Genetic_location	Ordered_location
Association Source -> Destination	PAGE-OM::Page	Genetic_location

7.1.6.11 Genetic_map

Type: Class_Ordered_map

Package: SNP

Document:

A map containing the order and non-metric distance between genetic markers for identifiable sequence features. Unit: centiMorgan.

Connections

Connector	Source	Target
Association Destination -> Source	Genetic_map	Genetic_location
Generalization	Genetic_map	Ordered_map
Association Source -> Destination	PAGE-OM::Page	Genetic_map

7.1.6.12 Genomic_reference_allele

Type: Class_Genomic_allele

Package: SNP

Document:

The DNA sequence of a Genomic_variation as it appears in the public database.

Issue 12999 - Attribute “one of” renamed to “one_of”

Connections

Connector	Source	Target
Generalization	Genomic_reference_allele	SNP2::Genomic_allele
Association Source -> Destination	Reference_genomic_assembly	present_in Genomic_reference_allele
Association Source -> Destination	one_of Genomic_reference_allele	SNP2::Genomic_allele
Association Source -> Destination	PAGE-OM::Page	Genomic_reference_allele

7.1.6.13 Geographic_location

Type: Class_Identifiable

Package: SNP

Document:

Location of an individual or population in a geographic map. Locations are expressed in decimal degrees. Northern latitudes (if north of the equator) and eastern longitudes (if east of GM) have positive values by convention.

Connections

Connector	Source	Target
Association Source -> Destination	SNP2::Abstract_population	Geographic_location
Association Source -> Destination	PAGE-OM::Page	Geographic_location
Generalization Source -> Destination	Geographic_location	Identifiable

Attributes

Attribute	Notes
max_latitude double	Upper bound of a geographical location expressed in degrees from the equator.

max_longitude double	Upper bound of a geographical location expressed in degrees from the Greenwich meridian.
min_latitude double	Lower bound of a geographical location expressed in degrees from the equator.
min_longitude double	Lower bound of a geographical location expressed in degrees from the Greenwich meridian.

7.1.6.14 Identifiable

Type: Class

Package: SNP

Document:

Anything with a unique identifier.

Connections

Connector	Source	Target
Generalization	Subject_descriptor	Identifiable
Generalization Source -> Destination	SNP2::Latent_genotype	Identifiable
Generalization Source -> Destination	PAGE::Hypothesis	Identifiable
Aggregation supporting_source Source -> Destination	Identifiable	PAGE::Evidence
Generalization Source -> Destination	PAGE::Plate	Identifiable
Generalization Source -> Destination	SNP2::Abstract_run	Identifiable
Association Source -> Destination	Identifiable	Annotation
Association Source -> Destination	Identifiable	BREF::Publication
Association Source -> Destination	Identifiable	Db_xref
Generalization	BREF::Bibref_scope	Identifiable

Generalization	BREF::Journal	Identifiable
Association Source -> Destination	Identifiable	BREF::Source
Generalization Source -> Destination	Gene_product_change	Identifiable
Generalization Source -> Destination	PAGE::Algorithm	Identifiable
Generalization Source -> Destination	FUGE::Ontology_source	Identifiable
Generalization Source -> Destination	SNP2::Abstract_value	Identifiable
Generalization Source -> Destination	FUGE::Ontology_term	Identifiable
Generalization Source -> Destination	SNP2::Genomic_observation	Identifiable
Generalization Source -> Destination	BREF::Provider	Identifiable
Generalization Source -> Destination	Location	Identifiable
Generalization Source -> Destination	Taxon	Identifiable
Generalization Source -> Destination	PAGE::Study	Identifiable
Generalization Source -> Destination	PAGE::Conclusion	Identifiable
Generalization Source -> Destination	Methodology	Identifiable
Generalization Source -> Destination	Db_xref	Identifiable
Generalization Source -> Destination	Map	Identifiable

Generalization Source -> Destination	Anatomic_location	Identifiable
Generalization Source -> Destination	SNP2::Abstract_observation_target	Identifiable
Generalization Source -> Destination	PAGE::Genotype_phenotype_correlation_experiment	Identifiable
Generalization Source -> Destination	PAGE::Observable_feature_category	Identifiable
Generalization Source -> Destination	SNP2::Abstract_observable_feature	Identifiable
Generalization Source -> Destination	Geographic_location	Identifiable
Generalization Source -> Destination	Locatable	Identifiable
Generalization Source -> Destination	PAGE::Observation_method	Identifiable

Attributes

Attribute	Notes
creation_date string	Date of creation of the object.
delete_date string	Date of deletion of the object.
lsid string	Life Science Identifier.
modify_date string	Date of last modification of the object.
name string	Name can be non unique. Display name

7.1.6.15 Locatable

Type: Class_Identifiable

Package: SNP

Document:

Abstract class for anything that can be placed on a sequence, i.e., can act as a sequence feature.

Connections

Connector	Source	Target
Generalization	SNP2::Genomic_gene_structure	Locatable
Generalization Source -> Destination	SNP2::Genomic_variation	Locatable
Association Locations Source -> Destination	Locatable	Location
Generalization Source -> Destination	Residue_change	Locatable
Generalization Source -> Destination	Sequence	Locatable
Generalization Source -> Destination	Locatable	Identifiable

7.1.6.16 Location

Type: Class_Identifiable

Package: SNP

Document:

A position in an abstract map.

Connections

Connector	Source	Target
Association Source -> Destination	Location	Location
Generalization	SNP2::Reference_genomic_location	Location
Association Destination -> Source	Map	Location
Generalization Source -> Destination	Ordered_location	Location

Association Locations Source -> Destination	Locatable	Location
Generalization Source -> Destination	Location	Identifiable

7.1.6.17 Map

Issue 12998 - attribute “name” is removed

Type: Class_Identifiable

Package: SNP

Document:

An abstract map.

Connections

Connector	Source	Target
Generalization Source -> Destination	Physical_map	Map
Association Destination -> Source	Map	Location
Generalization Source -> Destination	Ordered_map	Map
Generalization Source -> Destination	Map	Identifiable

7.1.6.18 Methodology

Type: Class_Identifiable

Package: SNP

Document:

Abstract class for a laboratory method or a computational protocol.

Connections

Connector	Source	Target
Generalization	SNP2::Variation_assay	Methodology
Generalization Source -> Destination	SNP2::Haplotype_derivation_methods	Methodology
Generalization Source -> Destination	Methodology	Identifiable

7.1.6.19 Oligo

Type: Class_Sequence

Package: SNP

Document:

A DNA oligonucleotide used for detection and assay of Genomic_variations in Variation_assays. Synonym: primer

Connections

Connector	Source	Target
Generalization	Oligo	Sequence
Association Source -> Destination	SNP2::Variation_assay	Oligo
Association Source -> Destination	PAGE-OM::Page	Oligo

7.1.6.20 Ordered_location

Type: Class_Location

Package: SNP

Document:

A location on a secondary map that gives ordering information. That map can be a genetic or a cytogenetic map.

Connections

Connector	Source	Target
Generalization Source -> Destination	Genetic_location	Ordered_location
Generalization Source -> Destination	Ordered_location	Location
Association Destination -> Source	Ordered_location	SNP2::Genomic_variation
Generalization	Cytogenetic_location	Ordered_location

Attributes

Attribute	Notes
position string	Location description

7.1.6.21 Ordered_map

Type: Class_Map

Package: SNP

Document:

A non-metric map category.

Connections

Connector	Source	Target
Generalization	Cytogenetic_map	Ordered_map
Generalization	Genetic_map	Ordered_map
Generalization Source -> Destination	Ordered_map	Map

7.1.6.22 Organization

Issue 12998 - attribute “name” is removed

Type: Class_Provider

Package: SNP

Document:

Affiliation of a group of unidentified persons.

Connections

Connector	Source	Target
Generalization	Organization	BREF::Provider
Association Source -> Destination	PAGE-OM::Page	Organization

7.1.6.23 Person

Type: Class_Provider

Package: SNP

Document:

An individual identified by name who is involved with creating or disseminating publications. Possible roles are author, editor, contributor, publisher, or applicant.

Connections

Connector	Source	Target
Generalization	Person	BREF::Provider
Association Destination -> Source	editor Person	BREF::Book
Association Source -> Destination	PAGE-OM::Page	Person
Association Source -> Destination	PAGE::Evidence	curator Curator of evidence Person

Attributes

Attribute	Notes
affiliation string	Name of the organization the person is affiliated with.
email string	Email address
et_al boolean	Indicator that there are more persons in the list of persons; used typically for the bibliographic reference that contains only an abbreviated author list. Last listed author should have et_al set to true.
first_name string	First name
mid_initials string	Initials of any subsequent personal names.
postal_address string	Full postal address
surname string	Last name.

7.1.6.24 Physical_map

Type: Class_Map

Package: SNP

Document:

Map of physical entities, e.g., clones, contigs.

Connections

Connector	Source	Target
Generalization Source -> Destination	Physical_map	Map
Generalization	SNP2::Haplotype_map	Physical_map
Generalization Source -> Destination	Sequence_map	Physical_map

7.1.6.25 Reference_genomic_assembly

Type: Class_Sequence_map

Package: SNP

Document:

Set of reference sequences (Reference_genomic_landmarks) with an algorithm to define the genome and its constituents.

Connections

Connector	Source	Target
Association Destination -> Source	Reference_genomic_assembly	SNP2::Reference_genomic_location
Association Unspecified	Reference_genomic_landmark	Reference_genomic_assembly
Association Destination -> Source	Reference_genomic_assembly	SNP2::Reference_genomic_location_in_assembly
Association Unspecified	Reference_genomic_assembly	Taxon
Generalization	Reference_genomic_assembly	Sequence_map
Association Source -> Destination	Reference_genomic_assembly	present_in Genomic_reference_allele
Association Source -> Destination	PAGE-OM::Page	Reference_genomic_assembly

7.1.6.26 Reference_genomic_landmark

Type: Class_Sequence

Package: SNP

Document:

An accessioned genomic sequence that can be unambiguously assigned to a location in an assembly based on information in public databases.

Connections

Connector	Source	Target
Association Unspecified	Reference_genomic_landmark	Reference_genomic_assembly
Association Destination -> Source	Reference_genomic_landmark	SNP2::Reference_genomic_location
Association Destination -> Source	SNP2::Genomic_variation	Reference_genomic_landmark

Generalization	Reference_genomic_landmark	Sequence
Association Source -> Destination	PAGE-OM::Page	Reference_genomic_landmark

7.1.6.27 Residue_change

Type: Class_Gene_product_change, Locatable

Package: SNP

Document:

Abstract superclass for classes describing residue changes in gene-related sequences.

Connections

Connector	Source	Target
Generalization	SNP2::Translation_change	Residue_change
Generalization	SNP2::Genic_variation	Residue_change
Generalization Source -> Destination	Residue_change	Gene_product_change
Generalization Source -> Destination	Residue_change	Locatable
Generalization Source -> Destination	SNP2::Transcription_change	Residue_change

Attributes

Attribute	Notes
changed_residue string	The new residue in the reference sequence.
original_residue string	The affected residue in the reference sequence.

7.1.6.28 Sequence

Type: Class_Locatable, Sequence_map

Package: SNP

Document:

Biological sequence. A sequence accessioned by a public database and associated with an accessioned taxon, a version number and a release date.

Connections

Connector	Source	Target
Generalization	Oligo	Sequence
Generalization	CDS	Sequence
Generalization	SNP2::Genomic_allele	Sequence
Generalization	Reference_genomic_landmark	Sequence
Generalization Source -> Destination	Exon	Sequence
Generalization Source -> Destination	Sequence	Locatable
Generalization Source -> Destination	Sequence	Sequence_map

Attributes

Attribute	Notes
molecule string	Indicates the alphabet of the sequence molecule (e.g., 'DNA,' 'RNA,' 'protein.')
sequence string	The residue sequence string.

7.1.6.29 Sequence_map

Type: Class_Physical_map

Package: SNP

Document:

A map composed of one or more biological sequences.

Connections

Connector	Source	Target
Generalization	Reference_genomic_assembly	Sequence_map
Generalization Source -> Destination	Sequence	Sequence_map
Generalization Source -> Destination	Sequence_map	Physical_map

7.1.6.30 Subject_descriptor

Type: Class_Identifiable

Package: SNP

Document:

Subject term and vocabulary name container.

Connections

Connector	Source	Target
Generalization	Subject_descriptor	Identifiable
Association Destination -> Source	subject_heading Subject_descriptor	BREF::Bibref_subject
Association Destination -> Source	code Subject_descriptor	BREF::Bibref_subject
Association Destination -> Source	keyword Subject_descriptor	BREF::Bibref_subject
Association Source -> Destination	PAGE-OM::Page	Subject_descriptor

Attributes

Attribute	Notes
term string	The keyword, code, or subject heading value string.

vocabulary_name string	Name of the controlled vocabulary (e.g., Sears List of Subject Headings (SEARS), Library of Congress Subject Headings (LCSH), or Medical Subject Headings (MeSH).

7.1.6.31 Taxon

Type: Class_Identifiable

Package: SNP

Document:

Taxonomic unit. The UID is typically a species TaxID from the Taxonomy Database (http://www.ncbi.nih.gov/Taxonomy/).

Connections

Connector	Source	Target
Association Source -> Destination	SNP2::Abstract_population	Taxon
Association Unspecified	Reference_genomic_assembly	Taxon
Association Source -> Destination	PAGE-OM::Page	Taxon
Generalization Source -> Destination	Taxon	Identifiable

Attributes

Attribute	Notes
rank string	Rank of the taxon in the hierarchy. Typically ‘species.’
scientific_name string	Scientific name, e.g., Homo sapiens.

7.1.7 BREF

Type: Package «XSDschema»

Package: PAGE-OM

Bib - *(Logical diagram)*

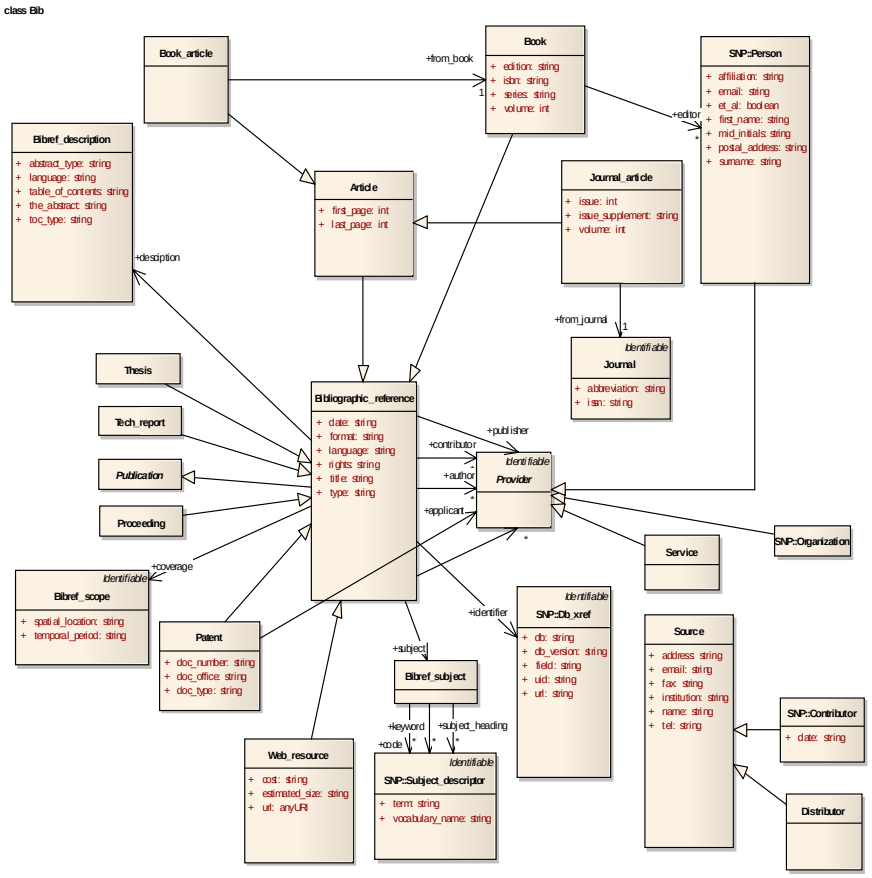

Figure 7.20

Issue 12998 - attribute “name” is removed

7.1.7.1 Article

Type: Class_Bibliographic_reference

Package: BREF

Document:

Non fictional prose forming an independent part of a publication.

Connections

Connector	Source	Target
Generalization	BREF::Journal_article	BREF::Article

Generalization	BREF::Article	BREF::Bibliographic_reference
Generalization	BREF::Book_article	BREF::Article

Attributes

Attribute	Notes
first_page int	First page in a article
last_page int	Last page in a article

7.1.7.2 Bibliographic_reference

Type: Class_Publication

Package: BREF

Document:

The BibliographicReference class is the core of the data model. It is a super-class for all specialized citation types, but it can also be instantiated and represent an additional type not specifically defined in this specification.

Connections

Connector	Source	Target
Generalization	BREF::Article	BREF::Bibliographic_reference
Generalization	BREF::Book	BREF::Bibliographic_reference
Generalization	BREF::Patent	BREF::Bibliographic_reference
Generalization	BREF::Thesis	BREF::Bibliographic_reference
Generalization	BREF::Web_resource	BREF::Bibliographic_reference
Association Destination -> Source	desciption BREF::Bibref_description	BREF::Bibliographic_reference

Generalization	BREF::Tech_report	BREF::Bibliographic_reference
Association Destination -> Source	status BREF::Entry_status	BREF::Bibliographic_reference
Generalization	BREF::Proceeding	BREF::Bibliographic_reference
Generalization	BREF::Bibliographic_reference	BREF::Publication
Association Destination -> Source	identifier Db_xref	BREF::Bibliographic_reference
Association Destination -> Source	publisher BREF::Provider	BREF::Bibliographic_reference
Association Destination -> Source	BREF::Provider	BREF::Bibliographic_reference
Association Destination -> Source	author BREF::Provider	BREF::Bibliographic_reference
Association Destination -> Source	subject BREF::Bibref_subject	BREF::Bibliographic_reference
Association Destination -> Source	coverage BREF::Bibref_scope	BREF::Bibliographic_reference
Association Destination -> Source	contributor BREF::Provider	BREF::Bibliographic_reference
Association Source -> Destination	PAGE-OM::Page	BREF::Bibliographic_reference

Attributes

Attribute	Notes
date string	Defines a date associated with an event in the life cycle of the cited resource, when this resource became available. Usually, it is a date of publishing, however, for not yet published resources, it can be a date of creation.
format string	Describes the physical or digital manifestation of the cited resource. It can have very different content depending on the citation type.

language string	Defines a language of the intellectual contents of the cited resource. The recommendation is to use values as defined by RFC1766 which includes a two-letter Language Code (taken from the ISO639 standard, followed optionally by a two-letter Country Code (taken from the ISO3166 standard). For example, “en” for English, “fr” for French, or “en-uk” for English used in the United Kingdom. Another possibility is to use MARC List of Languages.
rights string	Specifies information about rights over the cited resource. Typically, it contains a rights management statement for the resource, or it refers to a service providing such information. Rights information often encompasses Intellectual Property Rights [IPR], Copyrights, and various Property Rights.
title string	A title given to the cited resource (a name by which the resource is formally known).
type string	It defines the nature or genre of the cited resource. Although a working draft of Dublin Core Types recommends a type classification, the proposed types are mostly out of scope of this specification. The majority of cited resources would fall in the same category “text.” For the future, however, Dublin Core is considering the addition of subtypes to the high level types, or other ways of making sub-categories.

7.1.7.3 Bibref_description

Type: Class

Package: BREF

Document:

A brief account of the content of the cited resource. It can be either the abstract, or the table of contents, or both. It can be written in a language different from the language of the cited resource.

Connections

Connector	Source	Target
Association Destination -> Source	desciption BREF::Bibref_description	BREF::Bibliographic_reference
Association Source -> Destination	PAGE-OM::Page	BREF::Bibref_description

Attributes

Attribute	Notes
abstract_type string	Format of the abstract expressed in MIME (Multipurpose Internet Mail Extensions).
language string	Language of the description.
table_of_contents string	A list of divisions (chapters or articles) and the pages on which they start.
the_abstract string	Summary of the main points of the publication.

toc_type string	Format of the table of contents expressed in MIME (Multipurpose Internet Mail Extensions).

7.1.7.4 Bibref_scope

Type: Class_Identifiable

Package: BREF

Document:

It defines the extent or scope of the content of the cited resource. It can include spatial location (a place name or geographic co-ordinates), temporal period (a period label, date, or date range), or both. Finally, it can have additional dynamic properties such as jurisdiction).

Connections

Connector	Source	Target
Generalization	BREF::Bibref_scope	Identifiable
Association Destination -> Source	coverage BREF::Bibref_scope	BREF::Bibliographic_reference

Attributes

Attribute	Notes
spatial_location string	A place or area name or geographic coordinates in string format.
temporal_period string	A period label, date, or date range.

7.1.7.5 Bibref_subject

Type: Class

Package: BREF

Document:

It defines the topic of the content of the cited resource.

Connections

Connector	Source	Target
Association Destination -> Source	subject_heading Subject_descriptor	BREF::Bibref_subject

Association Destination -> Source	code Subject_descriptor	BREF::Bibref_subject
Association Destination -> Source	keyword Subject_descriptor	BREF::Bibref_subject
Association Destination -> Source	subject BREF::Bibref_subject	BREF::Bibliographic_reference

7.1.7.6 Book

Type: Class_Bibliographic_reference

Package: BREF

Document:

A written work or composition that has been published; usually printed on pages bound together.

Connections

Connector	Source	Target
Generalization	BREF::Book	BREF::Bibliographic_reference
Association Destination -> Source	from_book BREF::Book	BREF::Book_article
Association Destination -> Source	editor Person	BREF::Book

Attributes

Attribute	Notes
edition string	Name of the edition.
isbn string	International Standard Book Number
series string	Name of a collection of books.
volume int	Volume number of the book in a series.

7.1.7.7 Book_article

Type: Class_Article

Package: BREF

Document:

Article that forms part of a book.

Connections

Connector	Source	Target
Generalization	BREF::Book_article	BREF::Article
Association Destination -> Source	from_book BREF::Book	BREF::Book_article

7.1.7.8 Distributor

Type: Class_Source

Package: BREF

Document:

The original source of the data.

Connections

Connector	Source	Target
Generalization	BREF::Distributor	BREF::Source

7.1.7.9 Entry_status

Type: Class_Publication

Package: BREF

Document:

Defines information related to the citation itself rather than to the cited resource.

Connections

Connector	Source	Target
Association Destination -> Source	status BREF::Entry_status	BREF::Bibliographic_reference
Generalization	BREF::Entry_status	BREF::Publication

Attributes

Attribute	Notes
last_modified_date string	last date the citation entry was changed
subset string	Some bibliographic repositories consist of several, or even many, databases. The subset helps to locate the citation.

7.1.7.10 Journal

Type: Class_Identifiable

Package: BREF

Document:

A class describing a journal; a periodical dedicated to a particular subject. The citations referring to the journal articles have a reference to this class. There are only few explicit attributes defined, the rest are accessible using dynamic properties.

Connections

Connector	Source	Target
Generalization	BREF::Journal	Identifiable
Association Destination -> Source	from_journal BREF::Journal	BREF::Journal_article
Association Source -> Destination	PAGE-OM::Page	BREF::Journal

Issue 12998 - attribute “name” is removed

Attributes

Attribute	Notes
abbreviation string	Accepted abbreviated form of the journal name.
issn string	The ISSN (International Standard Serial Number) is an eight-digit number that identifies periodical publications as such.

7.1.7.11 Journal_article

Type: Class_Article

Package: BREF

Document:

Article, especially a scientific one that forms part of a journal.

Connections

Connector	Source	Target
Generalization	BREF::Journal_article	BREF::Article
Association Destination -> Source	from_journal BREF::Journal	BREF::Journal_article

Attributes

Attribute	Notes
issue int	Integer indicating the ordering of the issue within a volume.
issue_supplement string	Additional identifier for a separate supplement of the issue.
volume int	Volume number of the journal. Typically all issues of a journal published within a year belong to the same volume.

7.1.7.12 Patent

Type: Class_Bibliographic_reference

Package: BREF

Document:

A document granting an inventor sole rights to an invention.

Connections

Connector	Source	Target
Generalization	BREF::Patent	BREF::Bibliographic_reference
Association Destination -> Source	applicant BREF::Provider	BREF::Patent

Attributes

Attribute	Notes
doc_number string	Patent identifier
doc_office string	Name of the patent office (e.g., European Patent Office).
doc_type string	Type of a patent (e.g., ‘utility,’ ‘design,’ and ‘plant.’

7.1.7.13 Proceeding

Type: Class_Bibliographic_reference

Package: BREF

Document:

Printed communication from a scientific organization.

Connections

Connector	Source	Target
Generalization	BREF::Proceeding	BREF::Bibliographic_reference

7.1.7.14 Provider

Type: Class_Identifiable

Package: BREF

Document:

Bibliographic resources. The most obvious examples are authors, but it includes also publishers and other contributors.

These attributes define the active participants. They may be persons, organizations, or even services. A publisher is responsible for making the resource available. The authors and contributors are in ordered lists. The authors and contributors are responsible for creating the contents of the cited resource. There is no formal definition of how this 60 Single Nucleotide Polymorphisms Specification responsibility is divided between them. However, the authors are usually primary creators while contributors may be illustrators, translators, or other creative providers. Their role may be specified in a separate attribute in dynamic properties.

Connections

Connector	Source	Target
Generalization	Organization	BREF::Provider
Generalization	Person	BREF::Provider
Association Destination -> Source	publisher BREF::Provider	BREF::Bibliographic_reference
Association Destination -> Source	BREF::Provider	BREF::Bibliographic_reference
Association Destination -> Source	author BREF::Provider	BREF::Bibliographic_reference
Association Destination -> Source	applicant BREF::Provider	BREF::Patent
Association Destination -> Source	contributor BREF::Provider	BREF::Bibliographic_reference
Generalization Source -> Destination	BREF::Service	BREF::Provider
Generalization Source -> Destination	BREF::Provider	Identifiable

7.1.7.15 Publication

Type: Class

Package: BREF

Document:

A Citation. REF BibRef OMG standard. Not identifiable (Defined in model).

Connections

Connector	Source	Target
Association Source -> Destination	Identifiable	BREF::Publication
Generalization	BREF::Bibliographic_reference	BREF::Publication
Generalization	BREF::Entry_status	BREF::Publication

7.1.7.16 Service

Type: Class_Provider

Package: BREF

Document:

Provider of software service

Connections

Connector	Source	Target
Generalization Source -> Destination	BREF::Service	BREF::Provider
Association Source -> Destination	PAGE-OM::Page	BREF::Service

7.1.7.17 Source

Type: Class

Package: BREF

Document:

Source of the data. Individual or institute.

Connections

Connector	Source	Target
Generalization	BREF::Distributor	BREF::Source

Association Source -> Destination	Identifiable	BREF::Source
Generalization Source -> Destination	Contributor	BREF::Source
Association Source -> Destination	PAGE-OM::Page	BREF::Source

Attributes

Attribute	Notes
address string	Street address.
email string	Contact e-mail.
fax string	Fax number.
institution string	Name of the institution
name string	Name of the Source.
tel string	Telephone number.

7.1.7.18 Tech_report

Type: Class_Bibliographic_reference

Package: BREF

Document:

Technical report

Connections

Connector	Source	Target
Generalization	BREF::Tech_report	BREF::Bibliographic_reference

7.1.7.19 Thesis

Type: Class_Bibliographic_reference

Package: BREF

Document:

A treatise advancing a new point of view resulting from original research; usually a requirement for an advanced academic degree.

Connections

Connector	Source	Target
Generalization	BREF::Thesis	BREF::Bibliographic_reference

7.1.7.20 Web_resource

Type: Class_Bibliographic_reference

Package: BREF

Document:

A referred web address

Connections

Connector	Source	Target
Generalization	BREF::Web_resource	BREF::Bibliographic_reference

Attributes

Attribute	Notes
cost string	Cost of accessing the information
estimated_size string	Rough size of the retrieved reference entry
url anyURI	Uniform Resource Locator

8 Platform Specific Model

The previous chapter defines a platform independent model. The real implementations, however, are expected to depend on a more specific platform. This chapter shows an XML-based platform specific model expressing exchange format for data that conforms to platform independent model described above.

The platform specific model for XML derives its architecture according to the XML as suggested by W3C document "Extensible Markup Language (XML) 1.0 (http://www.w3.org/TR/REC-xml/)".

The model is defined by XML Schema as suggested by following W3C documents.

- XML Schema Part 0: Primer (http://www.w3.org/TR/xmlschema-0/)
- XML Schema Part 1: Structure (http://www.w3.org/TR/xmlschema-1/)
- XML Schema Part 2: Datatypes (http://www.w3.org/TR/xmlschema-2/)

The normative XML schema was generated from the modeling tool Enterprise Architect (EA), version 7.0.817.1. The target namespaces were propagated by the EA tool into the resulting XML Schema. However, because these target namespaces are not part of the XMI, when using a different tool, the namespaces must be changed in the resulting XML Schema manually as follows:

UML Package name	Target namespace
SNP	http://www.openpml.org/page-om/snp
SNP2	http://www.openpml.org/page-om/snp2
PAGE	http://www.openpml.org/page-om/page
BREF	http://www.openpml.org/page-om/bref
FUGE	http://www.openpml.org/page-om/fuge

Also following manual modifications were done into the XML schema files:
SchemaLocations attributes were added into the import elements for all packages (snp.xsd, snp2.xsd, page.xsd, bref.xsd and fuge.xsd) respectively.

Duplicate attributes coming from diamond inheritance are removed manually from snp.xsd file by editing out first occurrences of attributes from lsid to Source in Sequence and Residue elements.

The normative XML schema also uses consistently constructs `xs:choice minOccurs="0" maxOccurs="unbounded` instead of constructs `xs:sequence`.

The normative XML Schema is attached in the "Accompanied files" as file *PSM/PAGE-OM.xsd*.

Annex A
Accompanied Files

(Informative)

This annex is a set of the accompanied files. Some of these files are normative and some of them contain examples and convenient images. If there is a discrepancy between the contents of the normative files and this document, then the normative files take precedence.

The accompanied files are divided into two sections, PIM and PSM. The PIM section includes files related directly to the "Platform Independent Model," the PSM section contains the files used in the "Platform Specific Model."

Platform Independent Model

PAGE-OM_uml_2.1_xmi_2.1.xml

A normative representation of the PAGE-OM specification.

PAGE-OM.EAP

This is the main file created by the Enterprise Architect modeling tool. It served as a base for generating the XMI and description files, mentioned above.

BREF_Bib.png
BasicTypes.png
FUGE_Ontology.png
PAGE_Evidence_and_value.png
PAGE_Frequency.png
PAGE_Genotype.png
PAGE_Genotype2.png
PAGE_Identifiable.png
PAGE_Phenotype.png
PAGE_Phenotype2.png
Page_study.png
Root_bref.png
Root_snp.png
Root_snp2.png
SNP2_Haplotype.png
SNP2_Map.png
SNP2_Sample.png
SNP2_Sequence.png
SNP2_Sequence2.png
SNP_Map.png
SNP_Sample.png
SNP_Sequence.png
SNP_Sequence2.png

Diagrams of the PAGE-OM specification. These are the same diagrams as in this document but perhaps with a better resolution.

Platform Specific Model

all.xsd
basictypes.xsd
bref.xsd
fuge.xsd
page.xsd
snp.xsd
snp2.xsd

Annex B
References

(Informative)

- Sherry ST, Ward MH, Kholodov M, Baker J, Phan L, Smigielski EM, Sirotkin K. (2001) dbSNP: the NCBI database of genetic variation. Nucleic Acids Res. 29(1):308-311; see also http://www.ncbi.nlm.nih.gov/SNP/index.html
- Thorisson GA, Stein LD. (2003) The SNP Consortium website: past, present and future. Nucleic Acids Res. 31(1):124-127; see also http://snp.cshl.org/
- Fredman D, Siegfried M, Yuan YP, Bork P, Lehvaslaiho H, Brookes AJ. (2002) HGVbase: a human sequence variation database emphasizing data quality and a broad spectrum of data sources. Nucleic Acids Res. 30(1):387-391; see also http://www.hgvbase.org/
- Mika Hirakawa, Toshihiro Tanaka, Yoichi Hashimoto, Masako Kuroda, Toshihisa Takagi and Yusuke Nakamura (2002) JSNP: a database of common gene variations in the Japanese population. Nucleic Acids Res. 30(1):158-162; see also http://snp.ims.u-tokyo.ac.jp/
- Baker P G, Goble C A, Bechhofer S, Paton N W, Stevens R and Brass A (1999) An Ontology for Bioinformatics Applications. Bioinformatics 15, 510-520
- The Gene Ontology Consortium. Gene Ontology: tool for the unification of biology. Nature Genet. (2000) 25: 25-29.; see also http://www.geneontology.org/
- T.E. Klein, J.T. Chang, M.K. Cho, K.L. Easton, R. Fergerson, M. Hewett, Z. Lin, Y. Liu, S. Liu, D.E. Oliver, D.L. Rubin, F. Shafa, J.M. Stuart and R.B. Altman, "Integrating Genotype and Phenotype Information: An Overview of the PharmGKB Project", The Pharmacogenomics Journal (2001) 1, 167-170.; see also http://www.pharmgkb.org/
- Rajeevan H, Osier MV, Cheung KH, Deng H, Druskin L, Heinzen R, Kidd JR, Stein S, Pakstis AJ, Tosches NP, Yeh CC, Miller PL, Kidd KK. "ALFRED the ALlele FREquency Database update." Nucleic Acids Research..31(1):270-271.(2003); see also http://alfred.med.yale.edu/
- Mailman MD, Feolo M, Jin Y, Kimura M, Tryka K, Bagoutdinov R, Hao L, Kiang A, Paschall J, Phan L, Popova N, Pretel S, Ziyabari L, Lee M, Shao Y, Wang ZY, Sirotkin K, Ward M, Kholodov M, Zbicz K, Beck J, Kimelman M, Shevelev S, Preuss D, Yaschenko E, Graeff A, Ostell J, Sherry ST. The NCBI dbGaP database of genotypes and phenotypes. Nat Genet. 2007 Oct;39(10):1181-1186.
- Yamada Y, Kato K, Hibino T, Yokoi K, Matsuo H, Segawa T, Watanabe S, Ichihara S, Yoshida H, Satoh K, Nozawa Y. Prediction of genetic risk for metabolic syndrome. Atherosclerosis 2007;191:298-304.

Annex C
Glossary

(Informative)

Term	Definition
Anatomic location	Description of the anatomical location the molecular sample is derived from. Best expressed as a controlled vocabulary of anatomical terms.
Assay Set	Set of assays which are group experimentally. (e.g., microarray)
Assayed genomic genotype	The result of applying a Variation assay to an individual, to reveal one or more of the Genomic alleles carried by that individual. This term applies to the observed data rather than to the inferred state of the individual (see Consensus genomic genotype). Thus the same individual might have several different Assayed genomic genotypes at the same Genomic variation, where the variation might be due to differing assays, experimental error, dominant systems, missing data, and so forth. Synonym: Measurement.
Association Study	Examination of genetic variation across the human genome, designed to identify genetic associations with observable phenotypes
CDS	Region in a reference sequence that determines the start, end and strand of the gene coding region.
Consensus genomic genotype	A list of one or more Genomic alleles carried by a single individual for a single Genomic variation. Unlike the Assayed genomic genotype, the genomic genotype is intended to be used solely for assertions as to the true value of the alleles, after experimental error and other factors are taken into account. The number of Genomic alleles is equal to the ploidy (copy number) of the individual's genomic DNA at the Reference genomic location of the Genomic variation. Synonym: Inferred genomic genotype, Genomic genotype.
Contributor	Person who has edited data in the data set. Synonym: curator.
Cytogenetic location	A location in a cytogenetic map expressed as a band or a range of bands.
Cytogenetic map	Map of cytogenetic bands describing their relative order.
Db xref	Cross-reference between two Identifiable in different databases.
Disease	Special case of a phenotype describing a disease state.
Environment	Circumstances, objects, or conditions by which one is surrounded.

Exon	Region in a reference sequence that determines the start, end and strand of the transcript that is not spliced away.
Frequency Set	Set of frequencies for one genomic variation in one panel using one variation assay
Functional change	Change in the function of the final gene product.
Gene based haplotype	Collection of nucleotides (Genomic alleles in Genomic variations) that define a gene. In older usage same as locus.
Gene product change	Abstract superclass for a different genic change descriptors.
Genetic location	A relative location of a marker in a genetic map based on recombination.
Genetic map	A map containing the order and non-metric distance between genetic markers for identifiable sequence features. Unit: centi-morgan.
Genic variation	Genomic variation with location in coordinates. Synonym: mutation (when change from a common allele affects phenotype).
Genomic allele	One of several alternative DNA sequences of a Reference genomic location as it appears in the population of organisms. Synonym: variant, allele.
Genomic allele population frequency	The frequency with which a particular Genomic allele is seen in a particular Panel. This can be measured from pooled samples. Synonyms: Genomic allele panel frequency, allele frequency.
Genomic gene structure	A of a gene expressed as location of the CDS and exons. Defines genic coordinate system from start of the CDS downstream.
Genomic genotype population frequency	Frequency of a Consensus genomic genotype in a Panel.
Genomic haplotype	A set of Genomic alleles across an equal number of Genomic variations in a single chromosome and in a single individual. The Genomic haplotype is derived from a set of genomic genotype. For each Genomic variation, the haplotype contains one and only one Genomic allele. Furthermore, the Genomic alleles are required to be in phase on the individual, meaning that they are located on the same contiguous strand of DNA. Synonym: Haplotype.
Genomic haplotype population frequency	Frequency of a Genomic haplotype in a Panel.
Genomic reference allele	The DNA sequence of a Genomic variation as it appears in the public database.
Genomic Variation	A variable site in a Reference genomic landmark sequence. Synonyms: polymorphic site, marker
Genomic variation	A site in a Reference genomic sequence. Synonyms: polymorphic site, polymorphism, SNP.
Genotype Phenotype Correlation Experiment	Study to detect associations between genotype and phenotype. Can be family, population, or case/control based. Synonyms: Association study, Linkage Study.

Haplotype block	Large (a few kb to >100 kb) blocks of Genomic alleles in linkage disequilibrium (LD) and a few haplotypes per block, separated by region of recombination.
Haplotype derivation methods	Association class describing methods used to derive Genomic haplotypes from Consensus genomic genotypes.
Heterozygosity	A numerical measure of observed abundance of different Genomic alleles in a Genomic variation.
Haplotype map	Map of haplotypes. Features include: 1.
Individual	A single member of a species, where a species is an accessioned taxon defined by a public database, and the individual is accessioned in a public or private database. Synonym: "inbred strain" in homozygous lineages.
Methodology	Abstract class for a laboratory method or a computational protocol.
Molecular sample	A sample from Individual from a Panel defining the molecule and tissue/ cell used (Anatomic locations) in the Variation assay. Synonym: Sample of individual.
Neighbour variation	Another Genomic variation close enough to affect the Variation assay.
Observable Feature	Measurable feature of observable (e.g., size of nose)
Observable Feature Category	Observable category ideally expressed using an appropriate ontology
Observation Method	Method of observation (e.g., ruler). Can be also questionnaire.
Observation Target	Capable of being observed. For example nose size or eye color.
Observation Value	Observation done at specific point in time. Time is expressed according to the W3C profile version of an ISO 8601 date-time string. http:// www.w3.org/TR/NOTE-datetime
Oligo	A DNA oligo-nucleotide used for detection and assay of Genomic variations in Variation assay. Synonym: primer.
Ordered location	A location on a secondary map that gives ordering information. That map can be a genetic or a cytogenetic map.
Ordered map	A non-metric map category.
Panel	A set of samples from individuals drawn from the same species and used for genetic studies. A panel must be identifiable with a list of accessioned individuals, if possible. (Note: do not use "population" here!) Synonym: Sample Set, Sample from population(s). Plate in Coriel sense.
Phenotype	Observable part of the structure, function or behavior of a living organism.
Phenotype Disease Category	Category associated with disease phenotype. Best expressed using a disease ontology.
Physical map	Map of physical entities, e.g., clones, contigs.
Population	A interbreeding set of individuals, from whom a Panel is drawn.

Reference genomic assembly	Set of reference sequences (Reference genomic landmarks) with an algorithm to define the genome and its constituents.
Reference genomic landmark	An accessioned sequence which can be unambiguously assigned a location in an assembly based on information in public databases.
Reference genomic location	A location within a Reference genomic. Attributes of the location are the Reference genomic assembly and/or the Reference genomic landmark, the start and end range strand of the feature relative to the Reference genomic landmark.
Reference genomic location in assembly	A location in one chromosome of a reference genomic assembly. Instead of the reference sequence being an accessioned sequence, it is a versioned assembly.
Residue change	Abstract superclass for classes describing residue changes in gene-related sequences.
Sequence	Biological sequence. A sequence accessioned by a public database and associated with an accessioned taxon, a version number and a release date.
Sequence map	A map composed of one or more biological sequences.
Set of haplotypes	An abstract representation of a collection of haplotypes.
Structural change	Change in the 3D structure of the polypeptide chain.
Taxon	Taxonomic unit. The UID is typically a TaxID from the Taxonomy Database (http://www.ncbi.nih.gov/Taxonomy/).