Date: June 2005

Single Nucleotide Polymorphisms

OMG Available Specification

dtc/05-06-02

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Table of Contents

1 Scope

This response addresses the need for a standardized data exchange format for Single Nucleotide Polymorphisms (SNPs) entities in the Life Sciences domains. As a result of recent advances in SNPs research, a large amount of experimental data has been accumulating. Standardization is required to facilitate exchanging these data and developing interoperable systems for managing and analyzing them.

Single nucleotide polymorphisms (SNPs) are common DNA sequence variations that are the results of a single nucleotide changes in the genome sequence. SNPs have significant associations to variations in gene, amino acid, and protein. SNPs may be responsible for variation in phenotype or in disease. In SNPs database, information such as assay and publication is indispensable.

This specification models SNPs, using UML. The UML model, defining a platform independent model, describes the conceptual data structure. In order to actually perform data exchange, this specification formalizes a platform specific model, expressing the data exchange format in an XML Schema. The data exchange format is called PML, Polymorphisms Markup Language.

The typical use cases where the proposed data exchange format will be used include:

- Data upload or download between a public database and a local database, and
- an exchange (between researchers) of individual data items, consisting only from a very small subset of proposed XML elements and attributes. This is possible because of a "flat" character of the PML.

2 Conformance

The normative parts of this specification are:

- Platform independent model expressed in the attached XML file created according to XMI format rules, v1.2, using program Umbrello, version 1.4.
- Platform specific model, representing an XML-based data exchange format, defined by an XML Schema definition.

Any implementation using or producing given data exchange format is considered complying with this specification.

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.

3 Normative References

1. 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
2. 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/
3. 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://hgvbase.cgb.ki.se/
4. 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/
5. 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; see also http://cagpc.cs.man.ac.uk/tambis/manual/
6. The Gene Ontology Consortium (2001) Creating the gene ontology resource: design and implementation. Genome Res. 11:1425-1433; see also http://www.geneontology.org/
7. Hewett M, Oliver DE, Rubin DL, Easton KL, Stuart JM, Altman RB, Klein TE. (2002) PharmGKB: the Pharmacogenetics Knowledge Base. Nucleic Acids Res. 30(1):163-5; see also http://pharmgkb.org/
8. Cheung KH, Osier MV, Kidd JR, Pakstis AJ, Miller PL, Kidd KK. (2000) ALFRED: an allele frequency database for diverse populations and DNA polymorphisms. Nucleic Acids Res. 28(1):361-3; see also http://alfred.med.yale.edu/
9. Bibliographic query Service, an OMG specification, http://www.omg.org/technology/documents/formal/bibliographic_query.htm
10. Life Sciences Identifier, an OMG specification, http://www.omg.org/technology/documents/formal/life_sciences.htm

4 Terms and Definitions

The used terms are defined in Section 8.2, “Model classes, attributes and associations (details),” on page 14.

5 Symbols

This specification does not use any specific symbols/abbreviations.

6 Additional Information

6.1 Relationship to Existing OMG Specifications

- Bibliographic Query Service (BQS) - This specification uses attributes defined in BQS classes in order to describe publications. The relevant specification is available as OMG documents: formal/02-05-03, formal/02-05-04, and dtc/02-02-01.
- Life Sciences Identifiers (LSID) - There are many cross references in the model defined by this specification. They are expressed using LSIDs. The relevant specification is available as OMG documents: formal/04-12-01 and dtc/04-08-03.
- Genomic Map (GM) - The definitions in GM specification are either too limiting or too generic for SNPs purposes. The GM was designed at the time when genetic maps were the only genome wide maps. Current focus is on sequence maps.
- Model Driven Architecture (MDA) - MDA is used as a fundamental concept. Both platform-independent (PIM) and platform-specific model (PSM) are defined here.
- XML Interdata Interchange (XMI) - The platform-independent model for this specification for designed as a UML model that was converted into XMI model exchange format as defined in the XML Interdata Interchange specification.

6.2 Acknowledgements

The authors of this document wish to express their appreciation to those listed below (in alphabetic 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.

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7 Introduction

7.1 Overview

The graph below gives an overview how a genomic assembly (Reference_genomic_assembly) of sequences (Reference_genomic_landmark) is used to represent the genomic structure of a species (Taxon), and how known variable regions of the genome (Genomic_polymorphism) can be located (using Genomic_location) into one or more sequences and genomic assemblies.

Samples (Molecular_sample) from individuals (Individual) within the species (Taxon) are then used to assay (Polymorphism_assay) the variability (Genomic_alleles) within subpopulations (Panel) to gain knowledge of genotypes (Consensus_genomic_genotype) and haplotypes (Genomic_haplotype) and their relative abundance (see Frequency elsewhere in this document).

I

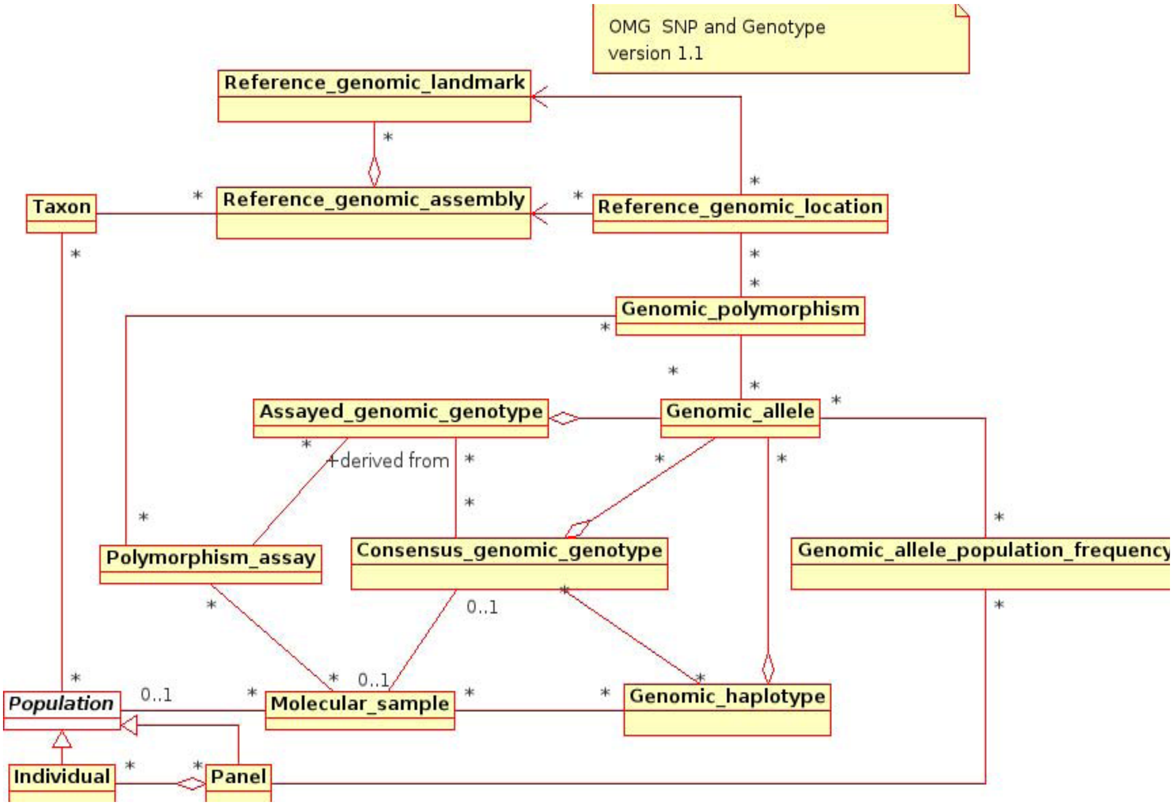


Figure 7.1

8 Platform Independent Model

The platform independent model is expressed as a UML model. The content of the model is explained below using the following techniques (note that independently on the techniques used here the normative expression of this model is the XMI-based attached XML file):

- The graphs showing subsets of the model. Each subset has its own short explanation. The same or similar explanation is also included in the XMI representation as description. The graphs may not show all attributes.
- The complete list of entities and their attributes, conveniently repeating also inherited attributes and associations.
- The words “derived from” are used in the UML model as comments, they do not imply any specific derivation algorithm.

8.1 Model subsets

8.1.1 Assay

An assay (Polymorphism_assay) is applied to a sample (Molecular_sample) to get a genotype (Assayed_genomic_genotype). Location of the sample can be described by several anatomical terms (Anatomic_location).

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_polymorhisms).

Genomic_haplotypes can sometimes be derived from genotypes.

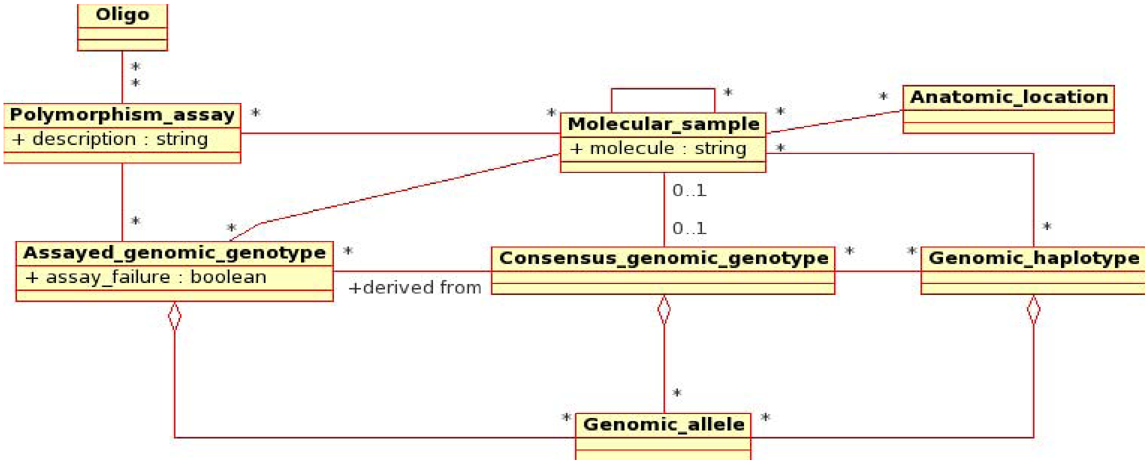


Figure 8.1

8.1.2 Individual

An Individual (Individual) interacts with the rest of the model by giving one or more samples (Molecular_samples), e.g at specific time points or from different tissues. A sample (Molecular_sample) can also belong to one or more population samples (Panel). Mixed samples and more complicated sample organizations are modeled by allowing Molecular_samples and Panels be composed of multiples of themselves. In these cases it is common that individuals cannot 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).

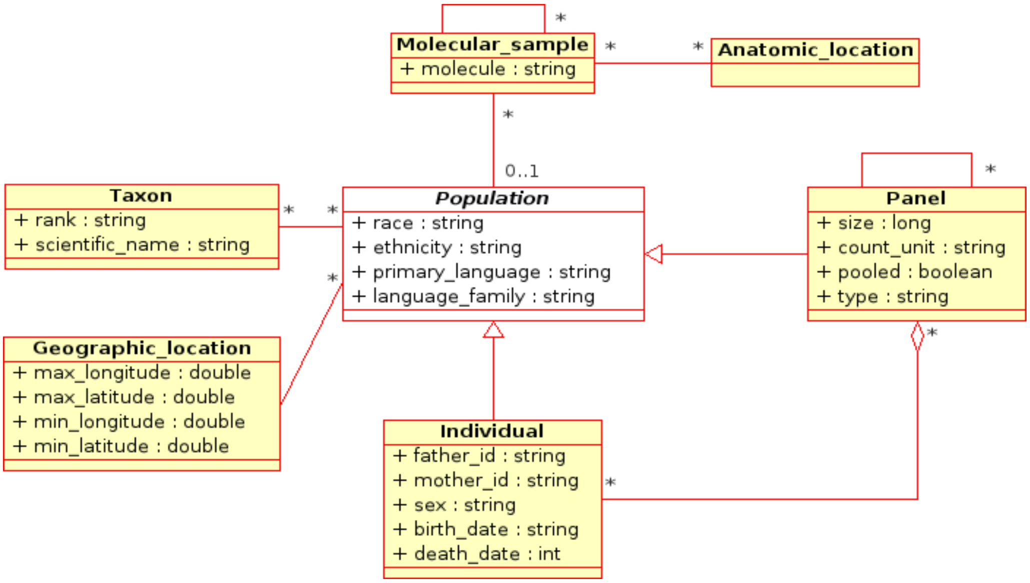


Figure 8.2

8.1.3 Frequency

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_polymorphism)in a sub-population (Panel).

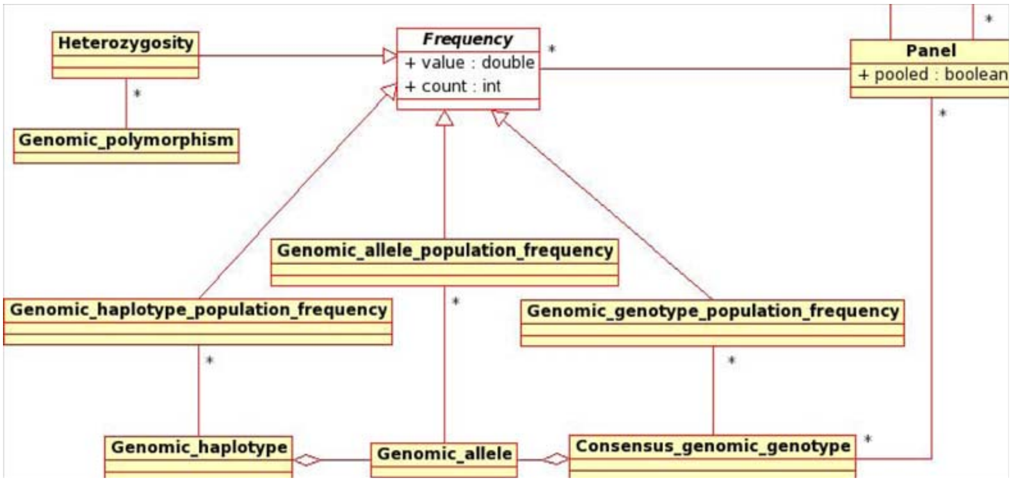


Figure 8.3

8.1.4 Haplotype

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).

Haplotypes and sets of haplotypes (Haplotype_block) can be placed on Haplotype_maps with additional information about their frequency and distribution.

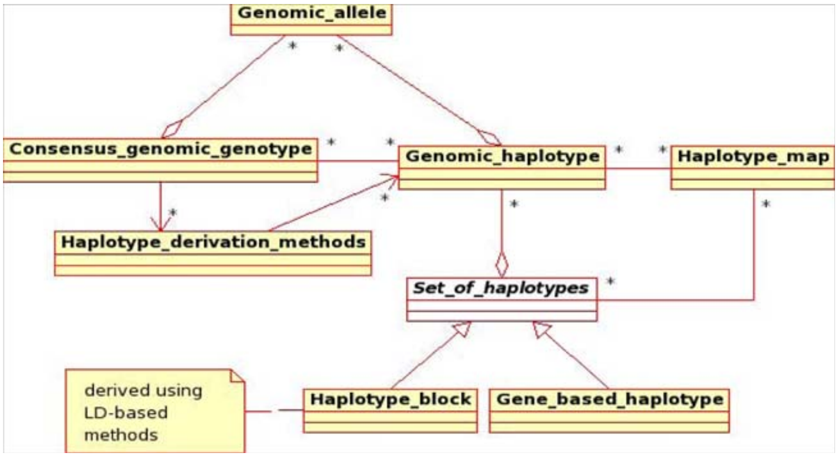


Figure 8.4

8.1.5 Map

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) which 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_polymorhism) 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 which 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).

8.1.6 Sequence

All sequences inherit from a generic Sequence class. All sequences (Sequence) and sequence features (e.g Genomic_polymorhism) can be located within a genomic sequence (Reference_genomic_sequence). This is modelled 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_polymorphisms are called Genomic_alleles. The genomic allele sequence present in the assembly (Reference_genomic_assembly) is labelled as Genomic_reference_allele. This together with the strong emphasis on having a position and especially strand in a genomic landmark for all polymorhisms are designed to help reducing the error from getting the orientation of the SNP wrong.

Sometimes polymorphisms, especially single nucleotide polymorphisms (SNPs), occur so close to each other that they affect each others detection oligo primers. It is useful to indicate these as Neighbor_variations.

Oligos (Oligo) are single strand DNA molecules used in polymorphism assays (Polymorphism_assay). It is important that the exact orientation ("strand") and location (Genomic_reference_location) of oligos are known so that the detected alleles (Genomic_alleles) can be identified unequivocally.

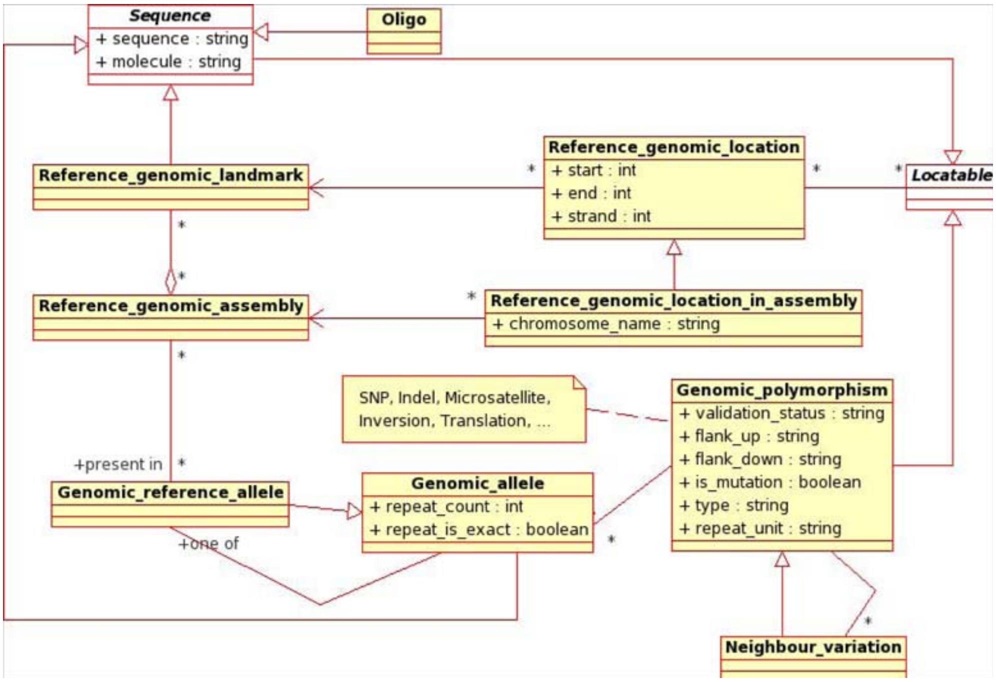


Figure 8.5

8.1.7 Translation

Polymorphisms (Genomic_polymorphisms) that occur with gene regions are of special interest. They (Genic_polymorphisms) can have an effect to the function of the gene. DNA changes that occur in low frequency in populations but that have a clear effect to the phenotype of an individual are commonly known as mutations. In this model mutations are Genomic_polymorphisms with attribute is_mutation set true.

The structure of the gene (Genomic_gene_structure) detailed enough to calculate the effects of mutations is composed of start, end and strand attributes of all the exons (Exon) and the coding region (CDS) locations (Reference_genomic_locations) in the reference sequence (Reference_genomic_landmark or Reference_genomic_assembly). Start of the gene defines then a new coordinate system with locations upstream getting negative locations. One Genomic_gene_structure here describes one transcript structure. More of these transcript structures are needed for genes with alternative transcripts. Genes located close to each other might be affected by the same polymorphism.

Any polymorphism (Genomic_polymorphism) can be understood to be part of the gene region. In practice, the region is commonly defined as 5000 nucleotide upstream from gene start to 5000 nucleotides downstream from the end of the coding region. This usually coves all regulatory regions of a gene.

A Genic_polymorphism can affect the transcript (have a Transcript_change) either directly by changing its nucleotide content or indirectly changing the amount of transcript produced. If the nucleotide content of the transcript is affected, it is customary to report the codon and its location in the coding region.

A Transcript_change can lead to a Translation_change if it affects the translated polypeptide. A Translation_change can have a known Functional_change either directly or via a Structural_change.

Any of these changes contain a description of the original state and changed (mutated) state. For sequence changes, part of the description is the residue change (e.g. A>T or M>I). An agreed ontology is needed to describe all changes. Examples of these are Sequence Ontology and the Mutation Event Controlled Vocabulary which is in use at EBI and built into Bioperl modules Bio::Variation.

There will undoubtedly be special cases where the relations between classes are variations of those presented in the model. For example, in the case of RNA genes the transcript change can have a Structural_change or Functional_change directly without Translation_change. These special cases should not be added into the model as they be easily added to the implementations and do not change the meaning of the classes.

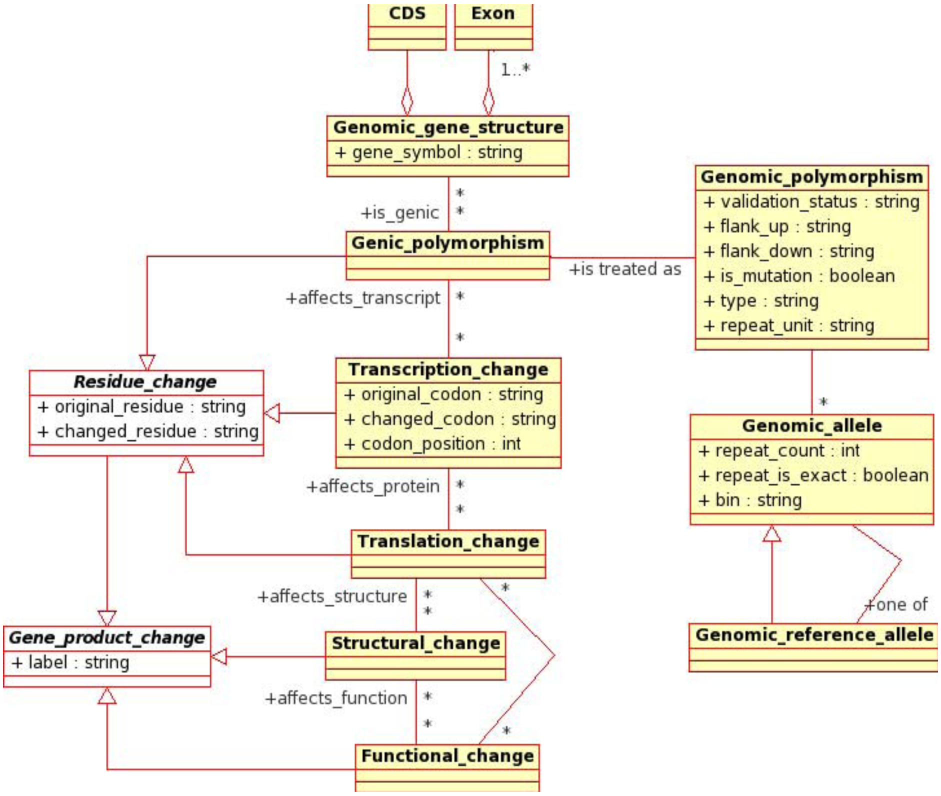
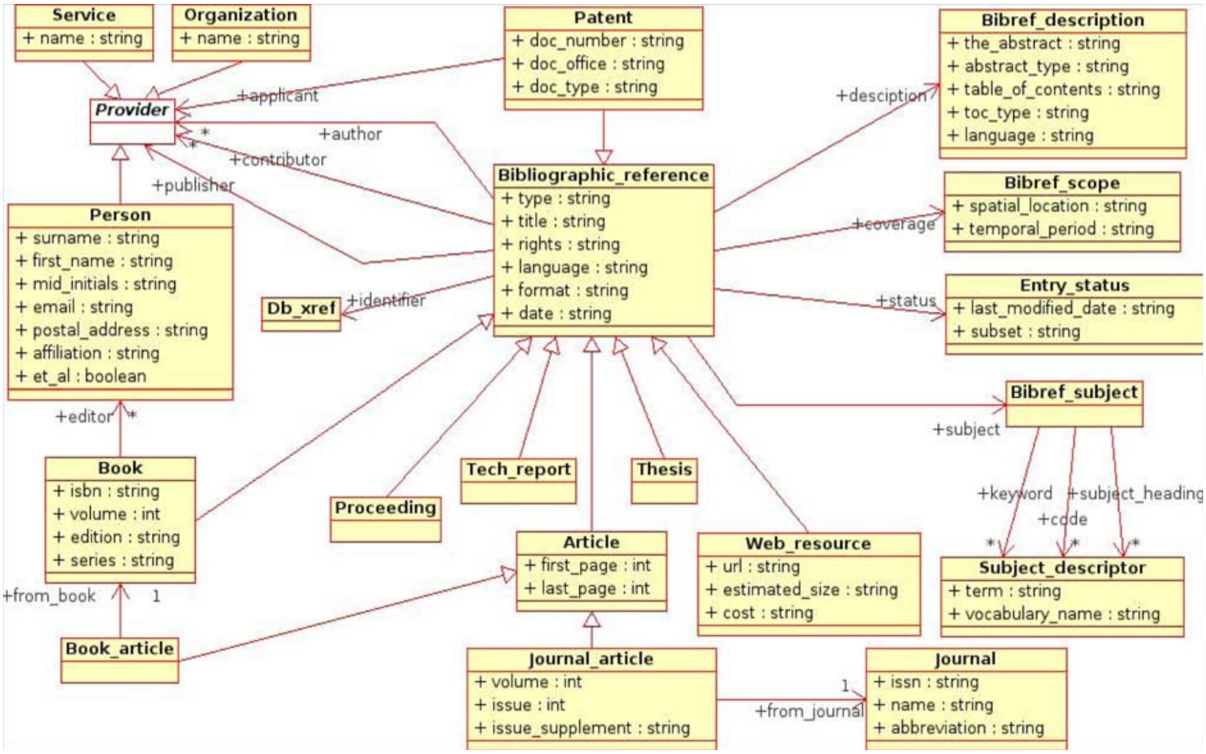


Figure 8.6

8.1.8 Bibliographic References

The Publication model is taken from the OMG Bibliographic Query Service. Together with usual attributes (described there, and formally expressed in the platform specific model here), such as "title" or "author", there is also an attribute "identifiable" containing a reference to any entities defined by this specification. The purpose is to allow chain-referencing (e.g., an entity A1 is linked to a publication P1 that is back linked to an entity B1 - so one can navigate from A1 to B1 via P1).



8.1.9 Identifiable

All classes in the model inherit from Identifiable. In this way, their instances are uniquely identifiable. Any Identifiable instance must have either attribute "id," or attribute "lsid." Usually, an "id" is used to identify an instance within a known context, and the "lsid" is used when cross-referencing to a different context. The "lsid" attribute follows syntax as defined in the OMG Life Sciences Identifiers 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.

Phenotype is included here as a place holder indicating that phenotype can link to many classes in the model but is really beyond the scope of this model.

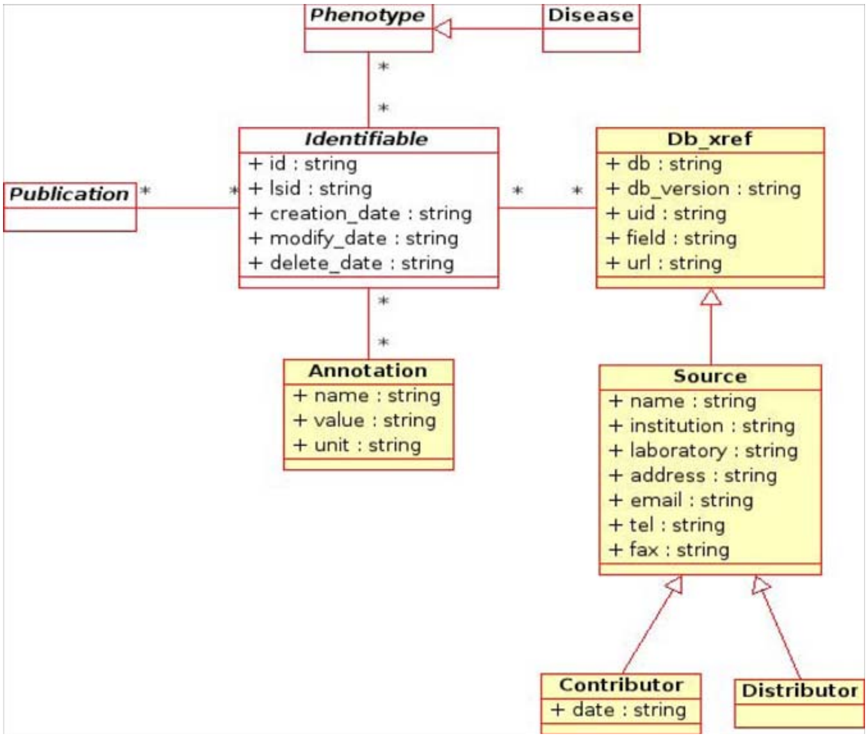


Figure 8.7

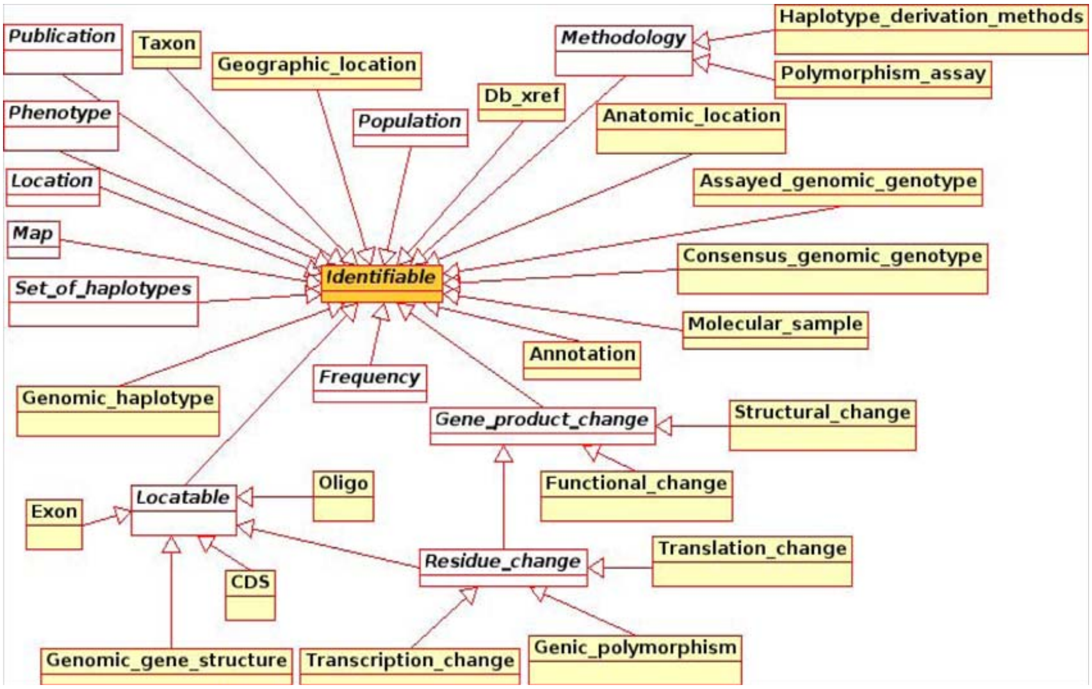


Figure 8.8

8.2 Model classes, attributes and associations (details)

Note that the details below can also be seen as an HTML document attached in the Accompanied Files.

8.2.1 Anatomic_location

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

Specialization of: Identifiable

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Molecular_sample** [defined at Anatomic_location]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.2 Annotation

A free text set of name, value, optional unit fields.

Specialization of: Identifiable

Attributes:

- **name** (string), [defined at Annotation] Name of the annotation feature.
- **value** (string), [defined at Annotation] Value assigned to the annotation feature.
- **unit** (string), [defined at Annotation] Optional unit of the annotation feature.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.

- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.3 Article

Nonfictional prose forming an independent part of a publication.

Specialization of: Bibliographic_reference

Attributes:

- **first_page** (int), [defined at Article] Page number of the first page.
- **last_page** (int), [defined at Article] Page number of the last page.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **type** (string), [defined at Bibliographic_reference] 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.
- **title** (string), [defined at Bibliographic_reference] A title given to the cited resource (a name by which the resource is formally known).
- **rights** (string), [defined at Bibliographic_reference] 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], Copyright, and various Property Rights.
- **language** (string), [defined at Bibliographic_reference] 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.
- **format** (string), [defined at Bibliographic_reference] Describes the physical or digital manifestation of the cited resource. It can have very different content depending on the citation type.
- **date** (string), [defined at Bibliographic_reference] 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.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Identifiable** [defined at Publication]
- to one **Bibref_description** [defined at Bibliographic_reference]
- to one **Bibref_subject** [defined at Bibliographic_reference]
- to one **Bibref_scope** [defined at Bibliographic_reference]
- to one **Entry_status** [defined at Bibliographic_reference]

8.2.4 Assayed_genomic_genotype

The result of applying a Polymorphism_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_polymorphism, where the variation might be due to differing assays, experimental error, dominant systems, missing data, and so forth. Synonym: Measurement

Specialization of: Identifiable

Attributes:

- **assay_failure** (boolean), [defined at Assayed_genomic_genotype] True if assay failed to result any genotype information.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Polymorphism_assay** [defined at Assayed_genomic_genotype]
- to one **Genomic_allele** [defined at Assayed_genomic_genotype]
- to one **Molecular_sample** [defined at Assayed_genomic_genotype]
- to one **Consensus_genomic_genotype** [defined at Assayed_genomic_genotype]
- to one **Publication** [defined at Identifiable]

- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.5 Bibliographic_reference

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.

Specialization of: Publication

Attributes:

- **type** (string), [defined at Bibliographic_reference] 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.
- **title** (string), [defined at Bibliographic_reference] A title given to the cited resource (a name by which the resource is formally known).
- **rights** (string), [defined at Bibliographic_reference] 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], Copyright, and various Property Rights.
- **language** (string), [defined at Bibliographic_reference] 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.
- **format** (string), [defined at Bibliographic_reference] Describes the physical or digital manifestation of the cited resource. It can have very different content depending on the citation type.
- **date** (string), [defined at Bibliographic_reference] 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.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Bibref_description** [defined at Bibliographic_reference]
- to one **Bibref_subject** [defined at Bibliographic_reference]

- to one **Bibref_scope** [defined at Bibliographic_reference]
- to one **Db_xref** [defined at Bibliographic_reference]
- to one **Entry_status** [defined at Bibliographic_reference]
- to one **Publication** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Identifiable** [defined at Publication]

8.2.6 Bibref_description

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

Attributes:

- **the_abstract** (string), [defined at Bibref_description] Summary of the main points of the publication.
- **abstract_type** (string), [defined at Bibref_description] Format of the abstract expressed in MIME (Multipurpose Internet Mail Extensions).
- **table_of_contents** (string), [defined at Bibref_description] A list of divisions (chapters or articles) and the pages on which they start.
- **toc_type** (string), [defined at Bibref_description] Format of the table of contents expressed in MIME (Multipurpose Internet Mail Extensions).
- **language** (string), [defined at Bibref_description] Language of the desciption

Associations:

- to one **Bibliographic_reference** [defined at Bibref_description]

8.2.7 Bibref_scope

It defines an 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).

Specialization of: Identifiable

Attributes:

- **spatial_location** (string), [defined at Bibref_scope] A place or area name or stringified geographic coordinates.
- **temporal_period** (string), [defined at Bibref_scope] A period label, date, or date range.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.

- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Bibliographic_reference** [defined at Bibref_scope]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.8 Bibref_subject

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

Attributes:

Associations:

- to one **Subject_descriptor** [defined at Bibref_subject]
- to one **Bibliographic_reference** [defined at Bibref_subject]

8.2.9 Book

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

Specialization of: Bibliographic_reference

Attributes:

- **isbn** (string), [defined at Book] International Standard Book Number
- **volume** (int), [defined at Book] Volume number of the book in a series.
- **edition** (string), [defined at Book] Name of the edition.
- **series** (string), [defined at Book] Name of a collection of books.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **type** (string), [defined at Bibliographic_reference] 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.

- **title** (string), [defined at Bibliographic_reference] A title given to the cited resource (a name by which the resource is formally known).
- **rights** (string), [defined at Bibliographic_reference] 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], Copyright, and various Property Rights.
- **language** (string), [defined at Bibliographic_reference] 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.
- **format** (string), [defined at Bibliographic_reference] Describes the physical or digital manifestation of the cited resource. It can have very different content depending on the citation type.
- **date** (string), [defined at Bibliographic_reference] 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.

Associations:

- to one **Book_article** [defined at Book]
- to one **Person** [defined at Book]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Identifiable** [defined at Publication]
- to one **Bibref_description** [defined at Bibliographic_reference]
- to one **Bibref_subject** [defined at Bibliographic_reference]
- to one **Bibref_scope** [defined at Bibliographic_reference]
- to one **Entry_status** [defined at Bibliographic_reference]

8.2.10 Book_article

Article that forms part of a book.

Specialization of: Article

Attributes:

- id (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.

- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **type** (string), [defined at Bibliographic_reference] 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.
- **title** (string), [defined at Bibliographic_reference] A title given to the cited resource (a name by which the resource is formally known).
- **rights** (string), [defined at Bibliographic_reference] 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], Copyright, and various Property Rights.
- **language** (string), [defined at Bibliographic_reference] 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.
- **format** (string), [defined at Bibliographic_reference] Describes the physical or digital manifestation of the cited resource. It can have very different content depending on the citation type.
- **date** (string), [defined at Bibliographic_reference] 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.
- **first_page** (int), [defined at Article] Page number of the first page.
- **last_page** (int), [defined at Article] Page number of the last page.

Associations:

- to one **Book** [defined at Book_article]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Identifiable** [defined at Publication]
- to one **Bibref_description** [defined at Bibliographic_reference]
- to one **Bibref_subject** [defined at Bibliographic_reference]
- to one **Bibref_scope** [defined at Bibliographic_reference]
- to one **Entry_status** [defined at Bibliographic_reference]

8.2.11 CDS

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

Specialization of: Locatable

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Genomic_gene_structure** [defined at CDS]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Reference_genomic_location** [defined at Locatable]

8.2.12 Consensus_genomic_genotype

A list of one or more Genomic_alleles carried by a single individual for a single Genomic_polymorphism. 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

Specialization of: Identifiable

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Genomic_allele** [defined at Consensus_genomic_genotype]

- to one **Panel** [defined at Consensus_genomic_genotype]
- to one **Assayed_genomic_genotype** [defined at Consensus_genomic_genotype]
- to one **Genomic_haplotype** [defined at Consensus_genomic_genotype]
- to one **Haplotype_derivation_methods** [defined at Consensus_genomic_genotype]
- to one **Molecular_sample** [defined at Consensus_genomic_genotype]
- to one **Genomic_genotype_population_frequency** [defined at Consensus_genomic_genotype]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.13 Contributor

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

Specialization of: Source

Attributes:

- **date** (string), [defined at Contributor] Date of the editing.
- **name** (string), [defined at Source] Name of the Source.
- **institution** (string), [defined at Source] The name of the institution.
- **laboratory** (string), [defined at Source] Laboratory or group within an institution.
- **address** (string), [defined at Source] Street address.
- **email** (string), [defined at Source] Contact email.
- **tel** (string), [defined at Source] Telephone number.
- **fax** (string), [defined at Source] FAX number.
- **db** (string), [defined at Db_xref] The remote database name or abbreviation.
- **db_version** (string), [defined at Db_xref] Version of the database
- **uid** (string), [defined at Db_xref] Unique identifier in the remote database.
- **field** (string), [defined at Db_xref] Field in the remote database where the UID value is found. E.g. ID or AC in EMBL. Optional.
- **url** (string), [defined at Db_xref] Full URL to the cross-refenced entry
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.

- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Bibliographic_reference** [defined at Db_xref]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.14 Cytogenetic_location

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

Specialization of: Ordered_location

Attributes:

- **position** (string), [defined at Ordered_location] Location description
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Cytogenetic_map** [defined at Cytogenetic_location]
- to one **Location** [defined at Location]
- to one **Map** [defined at Location]
- to one **Genomic_polymorphism** [defined at Ordered_location]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.15 Cytogenetic_map

Map of cytogenetic bands describing their relative order.

Specialization of: Ordered_map

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **name** (string), [defined at Map] Name of the map.
- **version** (double), [defined at Map] Version number of the map.

Associations:

- to one **Cytogenetic_location** [defined at Cytogenetic_map]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Location** [defined at Map]

8.2.16 Db_xref

Cross reference between two Identifiables in different databases.

Specialization of: Identifiable

Attributes:

- **db** (string), [defined at Db_xref] The remote database name or abbreviation.
- **db_version** (string), [defined at Db_xref] Version of the database
- **uid** (string), [defined at Db_xref] Unique identifier in the remote database.
- **field** (string), [defined at Db_xref] Field in the remote database where the UID value is found. E.g. ID or AC in EMBL. Optional.
- **url** (string), [defined at Db_xref] Full URL to the cross-refenced entry
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.

- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Bibliographic_reference** [defined at Db_xref]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.17 Disease

Special case of a phenotype describing a disease state.

Specialization of: Phenotype

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Identifiable** [defined at Phenotype]

8.2.18 Distributor

The original Source of the data.

Specialization of: Source

Attributes:

- **name** (string), [defined at Source] Name of the Source.

- **institution** (string), [defined at Source] The name of the institution.
- **laboratory** (string), [defined at Source] Laboratory or group within an institution.
- **address** (string), [defined at Source] Street address.
- **email** (string), [defined at Source] Contact email.
- **tel** (string), [defined at Source] Telephone number.
- **fax** (string), [defined at Source] FAX number.
- **db** (string), [defined at Db_xref] The remote database name or abbreviation.
- **db_version** (string), [defined at Db_xref] Version of the database
- **uid** (string), [defined at Db_xref] Unique identifier in the remote database.
- **field** (string), [defined at Db_xref] Field in the remote database where the UID value is found. E.g. ID or AC in EMBL. Optional.
- **url** (string), [defined at Db_xref] Full URL to the cross-refenced entry
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Bibliographic_reference** [defined at Db_xref]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.19 Entry_status

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

Specialization of: Identifiable

Attributes:

- **last_modified_date** (string), [defined at Entry_status] last date the citation entry was changed
- **subset** (string), [defined at Entry_status] Some bibliographic repositories consist of several, or even many, databases. The sub set helps to locate the citation.
- **id** (string), [defined at Identifiable] Unique identifier to the data.

- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Bibliographic_reference** [defined at Entry_status]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.20 Exon

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

Specialization of: Locatable

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Genomic_gene_structure** [defined at Exon]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Reference_genomic_location** [defined at Locatable]

8.2.21 Frequency

Abstract class for frequencies, expressed in percentages.

Abstract Class

Specialization of: Identifiable

Attributes:

- **value** (double), [defined at Frequency] Relative frequency value. Gets values between 0 and 1.
- **count** (int), [defined at Frequency] The actual count of items.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Panel** [defined at Identifiable]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.22 Functional_change

Change in the function of the final gene product.

Specialization of: Gene_product_change

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **label** (string), [defined at Gene_product_change] Human understandable term for the change. E.g. missense, truncation.

Associations:

- to one **Translation_change** [defined at Functional_change]
- to one **Structural_change** [defined at Functional_change]

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.23 Gene_based_haplotype

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

Specialization of: Set_of_haplotypes

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Haplotype_map** [defined at Set_of_haplotypes]
- to one **Genomic_haplotype** [defined at Set_of_haplotypes]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.24 Gene_product_change

Abstract superclass for a different genic change descriptors.

Abstract Class

Specialization of: Identifiable

Attributes:

- **label** (string), [defined at Gene_product_change] Human understandable term for the change. E.g. missense, truncation.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.

- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.25 Genetic_location

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

Specialization of: Ordered_location

Attributes:

- **position** (string), [defined at Ordered_location] Location description
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Genetic_map** [defined at Genetic_location]
- to one **Location** [defined at Location]
- to one **Map** [defined at Location]
- to one **Genomic_polymorphism** [defined at Ordered_location]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.26 Genetic_map

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

Specialization of: Ordered_map

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **name** (string), [defined at Map] Name of the map.
- **version** (double), [defined at Map] Version number of the map.

Associations:

- to one **Genetic_location** [defined at Genetic_map]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Location** [defined at Map]

8.2.27 Genic_polymorphism

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

Specialization of: Residue_change

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **original_residue** (string), [defined at Residue_change] The affected residue in the reference sequence.
- **changed_residue** (string), [defined at Residue_change] The new residue in the reference sequence.
- **label** (string), [defined at Gene_product_change] Human understandable term for the change. E.g. missense, truncation.

Associations:

- to one **Genomic_gene_structure** [defined at Genomic_polymorphism]
- to one **Genomic_polymorphism** [defined at Genomic_polymorphism]
- to one **Transcription_change** [defined at Genomic_polymorphism]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Reference_genomic_location** [defined at Locatable]

8.2.28 Genomic_allele

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

Specialization of: Sequence

Attributes:

- **repeat_count** (int), [defined at Genomic_allele] If the Genomic_variation type is 'microsatellite', get number of repeat units as value, e.g. 7.
- **repeat_is_exact** (boolean), [defined at Genomic_allele] If the Genomic_variation type is 'microsatellite', gets value true if the allele region consists of repeat units only.
- **bin** (string), [defined at Genomic_allele] Size class for microsatellite alleles when exact size can not be determined. Semantic name.
- **sequence** (string), [defined at Sequence] The residue sequence string.
- **molecule** (string), [defined at Sequence] Indicates the alphabet of the sequence molecule e.g. 'DNA', 'RNA', 'protein'
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **name** (string), [defined at Map] Name of the map.
- **version** (double), [defined at Map] Version number of the map.

Associations:

- to one **Consensus_genomic_genotype** [defined at Genomic_allele]

- to one **Genomic_polymorphism** [defined at Genomic_allele]
- to one **Assayed_genomic_genotype** [defined at Genomic_allele]
- to one **Genomic_haplotype** [defined at Genomic_allele]
- to one **Genomic_reference_allele** [defined at Genomic_allele]
- to one **Genomic_allele_population_frequency** [defined at Genomic_allele]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Reference_genomic_location** [defined at Locatable]
- to one **Location** [defined at Map]

8.2.29 Genomic_allele_population_frequency

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.

Specialization of: Frequency

Attributes:

- **value** (double), [defined at Frequency] Relative frequency value. Gets values between 0 and 1.
- **count** (int), [defined at Frequency] The actual count of items.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Genomic_allele** [defined at Genomic_allele_population_frequency]
- to one **Panel** [defined at Genomic_allele_population_frequency]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.30 Genomic_gene_structure

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

Specialization of: Locatable

Attributes:

- **gene_symbol** (string), [defined at Genomic_gene_structure] The gene symbol for the gene e.g. approved by the HUGO nomenclature committee.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Genic_polymorphism** [defined at Genomic_gene_structure]
- to one **Exon** [defined at Genomic_gene_structure]
- to one **CDS** [defined at Genomic_gene_structure]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Reference_genomic_location** [defined at Locatable]

8.2.31 Genomic_genotype_population_frequency

Frequency of a Consensus_genomic_genotype in a Panel.

Specialization of: Frequency

Attributes:

- **value** (double), [defined at Frequency] Relative frequency value. Gets values between 0 and 1.
- **count** (int), [defined at Frequency] The actual count of items.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.

- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Consensus_genomic_genotype** [defined at Genomic_genotype_population_frequency]
- to one **Panel** [defined at Frequency]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.32 Genomic_haplotype

A set of Genomic_alleles across an equal number of Genomic_polymorphisms in a single chromosome and in a single individual. The Genomic_haplotype is derived from a set of Consensus_genomic_genotype. For each Genomic_polymorphism, 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.

Specialization of: Identifiable

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Haplotype_derivation_methods** [defined at Genomic_haplotype]
- to one **Genomic_haplotype_population_frequency** [defined at Genomic_haplotype]
- to one **Genomic_allele** [defined at Genomic_haplotype]
- to one **Consensus_genomic_genotype** [defined at Genomic_haplotype]
- to one **Molecular_sample** [defined at Genomic_haplotype]
- to one **Haplotype_map** [defined at Genomic_haplotype]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]

- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.33 Genomic_haplotype_population_frequency

Frequency of a Genomic_haplotype in a Panel.

Specialization of: Frequency

Attributes:

- **value** (double), [defined at Frequency] Relative frequency value. Gets values between 0 and 1.
- **count** (int), [defined at Frequency] The actual count of items.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Genomic_haplotype** [defined at Genomic_haplotype_population_frequency]
- to one **Panel** [defined at Frequency]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.34 Genomic_polymorphism

A variable site in a Reference_genomic_landmark sequence. Synonyms: polymorphic_site, variation, SNP

Specialization of: Locatable

Attributes:

- **validation_status** (string), [defined at Genomic_polymorphism] Validation status, e.g. "Proven", "Suspected"
- **flank_up** (string), [defined at Genomic_polymorphism] Upstream flanking sequence (at least 25 residues, if possible).
- **flank_down** (string), [defined at Genomic_polymorphism] Downstream flanking sequence (at least 25 residues, if possible).
- **is_mutation** (boolean), [defined at Genomic_polymorphism] Proven phenotype change causing mutation.
- **type** (string), [defined at Genomic_polymorphism] The type of the polymorphism. E.g. SNP, microsatellite, indel,

translocation,...

- **repeat_unit** (string), [defined at Genomic_polymorphism] If type is 'microsatellite', gives the repeat unit, e.g. "CA"
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Heterozygosity** [defined at Genomic_polymorphism]
- to one **Genic_polymorphism** [defined at Genomic_polymorphism]
- to one **Reference_genomic_landmark** [defined at Genomic_polymorphism]
- to one **Neighbour_variation** [defined at Genomic_polymorphism]
- to one **Genomic_allele** [defined at Genomic_polymorphism]
- to one **Polymorphism_assay** [defined at Genomic_polymorphism]
- to one **Reference_genomic_location** [defined at Genomic_polymorphism]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.35 Genomic_reference_allele

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

Specialization of: Genomic_allele

Attributes:

- **repeat_count** (int), [defined at Genomic_allele] If the Genomic_variation type is 'microsatellite', get number of repeat units as value, e.g. 7.
- **repeat_is_exact** (boolean), [defined at Genomic_allele] If the Genomic_variation type is 'microsatellite', gets value true if the allele region consists of repeat units only.
- **bin** (string), [defined at Genomic_allele] Size class for microsatellite alleles when exact size can not be determined. Semantic name.
- **sequence** (string), [defined at Sequence] The residue sequence string.
- **molecule** (string), [defined at Sequence] Indicates the alphabet of the sequence molecule e.g. 'DNA', 'RNA', 'protein'

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **name** (string), [defined at Map] Name of the map.
- **version** (double), [defined at Map] Version number of the map.

Associations:

- to one **Genomic_allele** [defined at Genomic_reference_allele]
- to one **Reference_genomic_assembly** [defined at Genomic_reference_allele]
- to one **Consensus_genomic_genotype** [defined at Genomic_allele]
- to one **Genomic_polymorphism** [defined at Genomic_allele]
- to one **Assayed_genomic_genotype** [defined at Genomic_allele]
- to one **Genomic_haplotype** [defined at Genomic_allele]
- to one **Genomic_reference_allele** [defined at Genomic_allele]
- to one **Genomic_allele_population_frequency** [defined at Genomic_allele]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Reference_genomic_location** [defined at Locatable]
- to one **Location** [defined at Map]

8.2.36 Geographic_location

Location of an individual or population in a geographic map. Locations are expressed in decimal degrees. Northern latitides and eastern longitudes have positive values by convention.

Specialization of: Identifiable

Attributes:

- **max_longitude** (double), [defined at Geographic_location] Upper bound of a geographical location expressed in degrees from the Greenwich meridian.
- **max_latitude** (double), [defined at Geographic_location] Upper bound of a geographical location expressed in degrees from the equator.

- **min_longitude** (double), [defined at Geographic_location] Lower bound of a geographical location expressed in degrees from the Greenwich meridian.
- **min_latitude** (double), [defined at Geographic_location] Lower bound of a geographical location expressed in degrees from the equator.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.37 Haplotype_block

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.

Specialization of: Set_of_haplotypes

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Haplotype_map** [defined at Set_of_haplotypes]
- to one **Genomic_haplotype** [defined at Set_of_haplotypes]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.38 Haplotype_derivation_methods

Association class describing methods used to derive Genomic_haplotypes from Consensus_genomic_genotypes.

Specialization of: Methodology

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Consensus_genomic_genotype** [defined at Haplotype_derivation_methods]
- to one **Genomic_haplotype** [defined at Haplotype_derivation_methods]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.39 Haplotype_map

Map of haplotypes. Features include: * Block lenght 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

Specialization of: Physical_map

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **name** (string), [defined at Map] Name of the map.
- **version** (double), [defined at Map] Version number of the map.

Associations:

- to one **Genomic_haplotype** [defined at Haplotype_map]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Location** [defined at Map]

8.2.40 Heterozygosity

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

Specialization of: Frequency

Attributes:

- **value** (double), [defined at Frequency] Relative frequency value. Gets values between 0 and 1.
- **count** (int), [defined at Frequency] The actual count of items.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Genomic_polymorphism** [defined at Heterozygosity]
- to one **Panel** [defined at Frequency]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.41 Identifiable

Anything with a unique identifier.

Abstract Class

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.42 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.

Specialization of: Population

Attributes:

- **father_id** (string), [defined at Individual] Id of the father to allow building of pedigrees.
- **mother_id** (string), [defined at Individual] Id of the mother to allow building of pedigrees.
- **sex** (string), [defined at Individual] Recommended values are 'unknown', 'male' and 'female'. Additional values can be used to denote unusual karyotypes.
- **birth_date** (string), [defined at Individual] Date of birth of the individual. May be better abbreviated to birth year to protect the privacy of the individual.
- **death_date** (int), [defined at Individual] Date of death of the individual. May be better abbreviated to plain year to protect the privacy of the individual.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **race** (string), [defined at Population] Broad ethnic category of the population sample or "mixed".

- **ethnicity** (string), [defined at Population] Additional ethnic category of the population sample or "mixed".
- **primary_language** (string), [defined at Population] Language name or code, e.g. as in Ethnologue
- **language_family** (string), [defined at Population] Language family name or code, e.g. as in Ethnologue

Associations:

- to one **Panel** [defined at Individual]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Geographic_location** [defined at Population]
- to one **Taxon** [defined at Population]
- to one **Molecular_sample** [defined at Population]

8.2.43 Journal

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.

Specialization of: Identifiable

Attributes:

- **issn** (string), [defined at Journal] The ISSN (International Standard Serial Number) is an eight-digit number which identifies periodical publications as such.
- **name** (string), [defined at Journal] Full name
- **abbreviation** (string), [defined at Journal] Accepted abbreviated form of the journal name.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Journal_article** [defined at Journal]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]

- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.44 Journal_article

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

Specialization of: Article

Attributes:

- **volume** (int), [defined at Journal_article] Volume number of the journal. Typically all issues of a journal published within a year belong to a same volume.
- **issue** (int), [defined at Journal_article] Integer indicating the ordering of the issue within a volume.
- **issue_supplement** (string), [defined at Journal_article] Additional identifier for a separate supplement of the issue.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **type** (string), [defined at Bibliographic_reference] 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.
- **title** (string), [defined at Bibliographic_reference] A title given to the cited resource (a name by which the resource is formally known).
- **rights** (string), [defined at Bibliographic_reference] 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], Copyright, and various Property Rights.
- **language** (string), [defined at Bibliographic_reference] 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.
- **format** (string), [defined at Bibliographic_reference] Describes the physical or digital manifestation of the cited resource. It can have very different content depending on the citation type.
- **date** (string), [defined at Bibliographic_reference] 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.
- **first_page** (int), [defined at Article] Page number of the first page.
- **last_page** (int), [defined at Article] Page number of the last page.

Associations:

- to one **Journal** [defined at Journal_article]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Identifiable** [defined at Publication]
- to one **Bibref_description** [defined at Bibliographic_reference]
- to one **Bibref_subject** [defined at Bibliographic_reference]
- to one **Bibref_scope** [defined at Bibliographic_reference]
- to one **Entry_status** [defined at Bibliographic_reference]

8.2.45 Locatable

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

Abstract Class

Specialization of: Identifiable

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Reference_genomic_location** [defined at Locatable]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.46 Location

A position in an abstract map.

Abstract Class

Specialization of: Identifiable

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Location** [defined at Location]
- to one **Map** [defined at Location]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.47 Map

An abstract map.

Abstract Class

Specialization of: Identifiable

Attributes:

- **name** (string), [defined at Map] Name of the map.
- **version** (double), [defined at Map] Version number of the map.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Location** [defined at Map]

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.48 Methodology

Abstract class for a laboratory method or a computational protocol.

Abstract Class

Specialization of: Identifiable

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.49 Molecular_sample

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

Specialization of: Identifiable

Attributes:

- **molecule** (string), [defined at Molecular_sample] The molecule (RNA, DNA, protein) used in the assay.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.

- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Polymorphism_assay** [defined at Molecular_sample]
- to one **Anatomic_location** [defined at Molecular_sample]
- to one **Genomic_haplotype** [defined at Molecular_sample]
- to one **Molecular_sample** [defined at Molecular_sample]
- to one **Assayed_genomic_genotype** [defined at Molecular_sample]
- to one **Consensus_genomic_genotype** [defined at Molecular_sample]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.50 Neighbour_variation

Another Genomic_polymorphism close enough to affect the primer design.

Specialization of: Genomic_polymorphism

Attributes:

- **validation_status** (string), [defined at Genomic_polymorphism] Validation status, e.g. "Proven", "Suspected"
- **flank_up** (string), [defined at Genomic_polymorphism] Upstream flanking sequence (at least 25 residues, if possible).
- **flank_down** (string), [defined at Genomic_polymorphism] Downstream flanking sequence (at least 25 residues, if possible).
- **is_mutation** (boolean), [defined at Genomic_polymorphism] Proven phenotype change causing mutation.
- **type** (string), [defined at Genomic_polymorphism] The type of the polymorphism. E.g. SNP, microsatellite, indel, translocation,...
- **repeat_unit** (string), [defined at Genomic_polymorphism] If type is 'microsatellite', gives the repeat unit, e.g. "CA"
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Genomic_polymorphism** [defined at Neighbour_variation]
- to one **Heterozygosity** [defined at Genomic_polymorphism]
- to one **Genic_polymorphism** [defined at Genomic_polymorphism]
- to one **Reference_genomic_landmark** [defined at Genomic_polymorphism]
- to one **Neighbour_variation** [defined at Genomic_polymorphism]
- to one **Genomic_allele** [defined at Genomic_polymorphism]
- to one **Polymorphism_assay** [defined at Genomic_polymorphism]
- to one **Reference_genomic_location** [defined at Genomic_polymorphism]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.51 Oligo

A DNA oligonucleotide used for detection and assay of Genomic_polymorphisms in Polymorphism_assays. Synonym: primer

Specialization of: Sequence, Locatable

Attributes:

- **sequence** (string), [defined at Sequence] The residue sequence string.
- **molecule** (string), [defined at Sequence] Indicates the alphabet of the sequence molecule e.g. 'DNA', 'RNA', 'protein'
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **name** (string), [defined at Map] Name of the map.
- **version** (double), [defined at Map] Version number of the map.

Associations:

- to one **Polymorphism_assay** [defined at Oligo]
- to one **Publication** [defined at Identifiable]

- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Reference_genomic_location** [defined at Locatable]
- to one **Location** [defined at Map]

8.2.52 Ordered_location

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

Abstract Class

Specialization of: Location

Attributes:

- **position** (string), [defined at Ordered_location] Location description
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Genomic_polymorphism** [defined at Ordered_location]
- to one **Location** [defined at Location]
- to one **Map** [defined at Location]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.53 Ordered_map

A non-metric map category.

Abstract Class

Specialization of: Map

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **name** (string), [defined at Map] Name of the map.
- **version** (double), [defined at Map] Version number of the map.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Location** [defined at Map]

8.2.54 Organization

Affiliation of a group of unidentified persons.

Specialization of: Provider

Attributes:

- **name** (string), [defined at Organization] Full name of the organization
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]

- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Bibliographic_reference** [defined at Provider]
- to one **Patent** [defined at Provider]

8.2.55 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 term "population" here!) Synonym: SampleSet, Sample from population(s), "Plate" in Coriel sense.

Specialization of: Population

Attributes:

- **size** (long), [defined at Panel] The size of the sample. Note that the count_unit field affects how this value is interpreted.
- **count_unit** (string), [defined at Panel] Values are 'chromosome' and 'individual'. Default is 'individual'.
- **pooled** (boolean), [defined at Panel] True if accessioned individuals are not available.
- **type** (string), [defined at Panel] Optional identifier of the panel category: e.g plate, family, population sample
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **race** (string), [defined at Population] Broad ethnic category of the population sample or "mixed".
- **ethnicity** (string), [defined at Population] Additional ethnic category of the population sample or "mixed".
- **primary_language** (string), [defined at Population] Language name or code, e.g. as in Ethnologue
- **language_family** (string), [defined at Population] Language family name or code, e.g. as in Ethnologue

Associations:

- to one **Consensus_genomic_genotype** [defined at Panel]
- to one **Genomic_allele_population_frequency** [defined at Panel]
- to one **Panel** [defined at Panel]
- to one **Individual** [defined at Panel]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]

- to one **Phenotype** [defined at Identifiable]
- to one **Geographic_location** [defined at Population]
- to one **Taxon** [defined at Population]
- to one **Molecular_sample** [defined at Population]

8.2.56 Patent

A document granting an inventor sole rights to an invention.

Specialization of: Bibliographic_reference

Attributes:

- **doc_number** (string), [defined at Patent] Patent identifier
- **doc_office** (string), [defined at Patent] Name of the pattent office. E.g. European Patent Office
- **doc_type** (string), [defined at Patent] Type of a patent: e.g. 'utility', 'design', 'plant'
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **type** (string), [defined at Bibliographic_reference] 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.
- **title** (string), [defined at Bibliographic_reference] A title given to the cited resource (a name by which the resource is formally known).
- **rights** (string), [defined at Bibliographic_reference] 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], Copyright, and various Property Rights.
- **language** (string), [defined at Bibliographic_reference] 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.
- **format** (string), [defined at Bibliographic_reference] Describes the physical or digital manifestation of the cited resource. It can have very different content depending on the citation type.
- **date** (string), [defined at Bibliographic_reference] 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.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Identifiable** [defined at Publication]
- to one **Bibref_description** [defined at Bibliographic_reference]
- to one **Bibref_subject** [defined at Bibliographic_reference]
- to one **Bibref_scope** [defined at Bibliographic_reference]
- to one **Entry_status** [defined at Bibliographic_reference]

8.2.57 Person

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

Specialization of: Provider

Attributes:

- **surname** (string), [defined at Person] Last name.
- **first_name** (string), [defined at Person] First name
- **mid_initials** (string), [defined at Person] Initials of any subsequent personal names.
- **email** (string), [defined at Person] email address
- **postal_address** (string), [defined at Person] Full postal address
- **affiliation** (string), [defined at Person] Name of the organization the person is affiliated with.
- **et_al** (boolean), [defined at Person] Indicator that there are more persons in the list of persons; used typically the bibliographic reference contains only an abbrefiated author list. Last listed author should have et_al set to true.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Book** [defined at Person]
- to one **Publication** [defined at Identifiable]

- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Bibliographic_reference** [defined at Provider]
- to one **Patent** [defined at Provider]

8.2.58 Phenotype

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

Abstract Class

Specialization of: Identifiable

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Identifiable** [defined at Phenotype]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.59 Physical_map

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

Abstract Class

Specialization of: Map

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.

- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **name** (string), [defined at Map] Name of the map.
- **version** (double), [defined at Map] Version number of the map.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Location** [defined at Map]

8.2.60 Polymorphism_assay

An experimental protocol and set of reagents for detecting the Genomic_alleles of Genomic_polymorphisms carried by an individual or a panel of individuals. Synonym: Assay

Specialization of: Methodology

Attributes:

- **description** (string), [defined at Polymorphism_assay] Free text description of the assay protocol.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Genomic_polymorphism** [defined at Polymorphism_assay]
- to one **Molecular_sample** [defined at Polymorphism_assay]
- to one **Assayed_genomic_genotype** [defined at Polymorphism_assay]
- to one **Oligo** [defined at Polymorphism_assay]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.61 Population

A interbreeding set of individuals, from whom a Panel is drawn.

Abstract Class

Specialization of: Identifiable

Attributes:

- **race** (string), [defined at Population] Broad ethnic category of the population sample or "mixed".
- **ethnicity** (string), [defined at Population] Additional ethnic category of the population sample or "mixed".
- **primary_language** (string), [defined at Population] Language name or code, e.g. as in Ethnologue
- **language_family** (string), [defined at Population] Language family name or code, e.g. as in Ethnologue
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Geographic_location** [defined at Population]
- to one **Taxon** [defined at Population]
- to one **Molecular_sample** [defined at Population]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.62 Proceeding

Printed communication from an scientific organisation.

Specialization of: Bibliographic_reference

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.

- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **type** (string), [defined at Bibliographic_reference] 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.
- **title** (string), [defined at Bibliographic_reference] A title given to the cited resource (a name by which the resource is formally known).
- **rights** (string), [defined at Bibliographic_reference] 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], Copyright, and various Property Rights.
- **language** (string), [defined at Bibliographic_reference] 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.
- **format** (string), [defined at Bibliographic_reference] Describes the physical or digital manifestation of the cited resource. It can have very different content depending on the citation type.
- **date** (string), [defined at Bibliographic_reference] 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.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Identifiable** [defined at Publication]
- to one **Bibref_description** [defined at Bibliographic_reference]
- to one **Bibref_subject** [defined at Bibliographic_reference]
- to one **Bibref_scope** [defined at Bibliographic_reference]
- to one **Entry_status** [defined at Bibliographic_reference]

8.2.63 *Provider*

The class Provider and its sub-classes define active participants of the process of creation and dissemination of the 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 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.

Abstract Class

Specialization of: Identifiable

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Bibliographic_reference** [defined at Provider]
- to one **Patent** [defined at Provider]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.64 Publication

A Citation.

Abstract Class

Specialization of: Identifiable

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Identifiable** [defined at Publication]

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.65 Reference_genomic_assembly

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

Specialization of: Sequence_map

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **name** (string), [defined at Map] Name of the map.
- **version** (double), [defined at Map] Version number of the map.

Associations:

- to one **Reference_genomic_location** [defined at Reference_genomic_assembly]
- to one **Genomic_reference_allele** [defined at Reference_genomic_assembly]
- to one **Reference_genomic_landmark** [defined at Reference_genomic_assembly]
- to one **Taxon** [defined at Reference_genomic_assembly]
- to one **Reference_genomic_location_in_assembly** [defined at Reference_genomic_assembly]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Location** [defined at Map]

8.2.66 Reference_genomic_landmark

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

Specialization of: Sequence

Attributes:

- **sequence** (string), [defined at Sequence] The residue sequence string.
- **molecule** (string), [defined at Sequence] Indicates the alphabet of the sequence molecule e.g. 'DNA', 'RNA', 'protein'
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **name** (string), [defined at Map] Name of the map.
- **version** (double), [defined at Map] Version number of the map.

Associations:

- to one **Reference_genomic_assembly** [defined at Reference_genomic_landmark]
- to one **Genomic_polymorphism** [defined at Reference_genomic_landmark]
- to one **Reference_genomic_location** [defined at Reference_genomic_landmark]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Location** [defined at Map]

8.2.67 Reference_genomic_location

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.

Specialization of: Location

Attributes:

- **start** (int), [defined at Reference_genomic_location] Start of the location in the reference sequence.
- **end** (int), [defined at Reference_genomic_location] End of the location in the reference sequence.
- **strand** (string), [defined at Reference_genomic_location] Orientation of the feature in the reference sequence. One of 'forward', 'reverse', 'unknown'. Deafults to 'unknown'.
- **id** (string), [defined at Identifiable] Unique identifier to the data.

- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Reference_genomic_assembly** [defined at Reference_genomic_location]
- to one **Reference_genomic_landmark** [defined at Reference_genomic_location]
- to one **Genomic_polymorphism** [defined at Reference_genomic_location]
- to one **Location** [defined at Location]
- to one **Map** [defined at Location]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.68 Reference_genomic_location_in_assembly

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

Specialization of: Reference_genomic_location

Attributes:

- **chromosome_name** (string), [defined at Reference_genomic_location_in_assembly] Name of the chromosome in the assembly.
- **start** (int), [defined at Reference_genomic_location] Start of the location in the reference sequence.
- **end** (int), [defined at Reference_genomic_location] End of the location in the reference sequence.
- **strand** (string), [defined at Reference_genomic_location] Orientation of the feature in the reference sequence. One of 'forward', 'reverse', 'unknown'. Deafults to 'unknown'.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Reference_genomic_assembly** [defined at Reference_genomic_location_in_assembly]
- to one **Reference_genomic_landmark** [defined at Reference_genomic_location]
- to one **Genomic_polymorphism** [defined at Reference_genomic_location]
- to one **Location** [defined at Location]
- to one **Map** [defined at Location]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.69 Residue_change

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

Abstract Class

Specialization of: Locatable, Gene_product_change

Attributes:

- **original_residue** (string), [defined at Residue_change] The affected residue in the reference sequence.
- **changed_residue** (string), [defined at Residue_change] The new residue in the reference sequence.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **label** (string), [defined at Gene_product_change] Human understandable term for the change. E.g. missense, truncation.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Reference_genomic_location** [defined at Locatable]

8.2.70 Sequence

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

Abstract Class

Specialization of: Sequence_map, Locatable

Attributes:

- **sequence** (string), [defined at Sequence] The residue sequence string.
- **molecule** (string), [defined at Sequence] Indicates the alphabet of the sequence molecule e.g. 'DNA', 'RNA', 'protein'
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **name** (string), [defined at Map] Name of the map.
- **version** (double), [defined at Map] Version number of the map.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Reference_genomic_location** [defined at Locatable]
- to one **Location** [defined at Map]

8.2.71 Sequence_map

A map composed of one or more biological sequences.

Abstract Class

Specialization of: Physical_map

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.

- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **name** (string), [defined at Map] Name of the map.
- **version** (double), [defined at Map] Version number of the map.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Location** [defined at Map]

8.2.72 Service

Provide of software service

Specialization of: Provider

Attributes:

- **name** (string), [defined at Service] Full name.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Bibliographic_reference** [defined at Provider]
- to one **Patent** [defined at Provider]

8.2.73 Set_of_haplotypes

An abstact representation of a collection of haplotypes.

Abstract Class

Specialization of: Identifiable

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Haplotype_map** [defined at Set_of_haplotypes]
- to one **Genomic_haplotype** [defined at Set_of_haplotypes]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.74 Source

Source of the data. Individual or institute.

Specialization of: Db_xref

Attributes:

- **name** (string), [defined at Source] Name of the Source.
- **institution** (string), [defined at Source] The name of the institution.
- **laboratory** (string), [defined at Source] Laboratory or group within an institution.
- **address** (string), [defined at Source] Street address.
- **email** (string), [defined at Source] Contact email.
- **tel** (string), [defined at Source] Telephone number.
- **fax** (string), [defined at Source] FAX number.
- **db** (string), [defined at Db_xref] The remote database name or abbreviation.
- **db_version** (string), [defined at Db_xref] Version of the database
- **uid** (string), [defined at Db_xref] Unique identifier in the remote database.

- **field** (string), [defined at Db_xref] Field in the remote database where the UID value is found. E.g. ID or AC in EMBL. Optional.
- **url** (string), [defined at Db_xref] Full URL to the cross-refenced entry
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Bibliographic_reference** [defined at Db_xref]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.75 Structural_change

Change in the 3D structure of the polypeptide chain.

Specialization of: Gene_product_change

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **label** (string), [defined at Gene_product_change] Human understandable term for the change. E.g. missense, truncation.

Associations:

- to one **Translation_change** [defined at Structural_change]
- to one **Functional_change** [defined at Structural_change]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]

- to one **Phenotype** [defined at Identifiable]

8.2.76 Subject_descriptor

Subject term and vocabulary name container.

Specialization of: Identifiable

Attributes:

- **term** (string), [defined at Subject_descriptor] The keyword, code or subject heading value string
- **vocabulary_name** (string), [defined at Subject_descriptor] Name of the controlled vocabulary. E.g. Sears List of Subject Headings (SEARS), Library of Congress Subject Headings (LCSH), or Medical Subject Headings (MeSH).
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Bibref_subject** [defined at Subject_descriptor]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.77 Taxon

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

Specialization of: Identifiable

Attributes:

- **rank** (string), [defined at Taxon] Rank of the taxon in the hierarchy. Typically 'species'.
- **scientific_name** (string), [defined at Taxon] Scientific name, e.g. Homo sapiens.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.

- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.

Associations:

- to one **Reference_genomic_assembly** [defined at Taxon]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]

8.2.78 Tech_report

Technical report

Specialization of: Bibliographic_reference

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **type** (string), [defined at Bibliographic_reference] 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.
- **title** (string), [defined at Bibliographic_reference] A title given to the cited resource (a name by which the resource is formally known).
- **rights** (string), [defined at Bibliographic_reference] 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], Copyright, and various Property Rights.
- **language** (string), [defined at Bibliographic_reference] 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.
- **format** (string), [defined at Bibliographic_reference] Describes the physical or digital manifestation of the cited resource. It can have very different content depending on the citation type.
- **date** (string), [defined at Bibliographic_reference] 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.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Identifiable** [defined at Publication]
- to one **Bibref_description** [defined at Bibliographic_reference]
- to one **Bibref_subject** [defined at Bibliographic_reference]
- to one **Bibref_scope** [defined at Bibliographic_reference]
- to one **Entry_status** [defined at Bibliographic_reference]

8.2.79 Thesis

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

Specialization of: Bibliographic_reference

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **type** (string), [defined at Bibliographic_reference] 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.
- **title** (string), [defined at Bibliographic_reference] A title given to the cited resource (a name by which the resource is formally known).
- **rights** (string), [defined at Bibliographic_reference] 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], Copyright, and various Property Rights.
- **language** (string), [defined at Bibliographic_reference] 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.
- **format** (string), [defined at Bibliographic_reference] Describes the physical or digital manifestation of the cited resource.

It can have very different content depending on the citation type.

- **date** (string), [defined at Bibliographic_reference] 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.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Identifiable** [defined at Publication]
- to one **Bibref_description** [defined at Bibliographic_reference]
- to one **Bibref_subject** [defined at Bibliographic_reference]
- to one **Bibref_scope** [defined at Bibliographic_reference]
- to one **Entry_status** [defined at Bibliographic_reference]

8.2.80 Transcription_change

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

Specialization of: Residue_change

Attributes:

- **original_codon** (string), [defined at Transcription_change] The affected codon in the transcript.
- **changed_codon** (string), [defined at Transcription_change] The new codon in the transcript, if applicable.
- **codon_position** (int), [defined at Transcription_change] The first affected nucleotide in the codon. Values are: 1, 2 or 3.
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **original_residue** (string), [defined at Residue_change] The affected residue in the reference sequence.
- **changed_residue** (string), [defined at Residue_change] The new residue in the reference sequence.
- **label** (string), [defined at Gene_product_change] Human understandable term for the change. E.g. missense, truncation.

Associations:

- to one **Translation_change** [defined at Transcription_change]
- to one **Genic_polymorphism** [defined at Transcription_change]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Reference_genomic_location** [defined at Locatable]

8.2.81 Translation_change

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

Specialization of: Residue_change

Attributes:

- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **original_residue** (string), [defined at Residue_change] The affected residue in the reference sequence.
- **changed_residue** (string), [defined at Residue_change] The new residue in the reference sequence.
- **label** (string), [defined at Gene_product_change] Human understandable term for the change. E.g. missense, truncation.

Associations:

- to one **Transcription_change** [defined at Translation_change]
- to one **Structural_change** [defined at Translation_change]
- to one **Functional_change** [defined at Translation_change]
- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]
- to one **Phenotype** [defined at Identifiable]
- to one **Reference_genomic_location** [defined at Locatable]

8.2.82 Web_resource

A refereed web address

Specialization of: Bibliographic_reference

Attributes:

- **url** (string), [defined at Web_resource] Uniform Resource Locator
- **estimated_size** (string), [defined at Web_resource] Rought size of the retrieved reference entry.
- **cost** (string), [defined at Web_resource] Cost of accessing the infromation
- **id** (string), [defined at Identifiable] Unique identifier to the data.
- **lsid** (string), [defined at Identifiable] Life Science IDentifier.
- **creation_date** (string), [defined at Identifiable] Date of creation of the object.
- **modify_date** (string), [defined at Identifiable] Date of last modification of the object.
- **delete_date** (string), [defined at Identifiable] Date of deletion of the object.
- **type** (string), [defined at Bibliographic_reference] 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.
- **title** (string), [defined at Bibliographic_reference] A title given to the cited resource (a name by which the resource is formally known).
- **rights** (string), [defined at Bibliographic_reference] 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], Copyright, and various Property Rights.
- **language** (string), [defined at Bibliographic_reference] 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.
- **format** (string), [defined at Bibliographic_reference] Describes the physical or digital manifestation of the cited resource. It can have very different content depending on the citation type.
- **date** (string), [defined at Bibliographic_reference] 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.

Associations:

- to one **Publication** [defined at Identifiable]
- to one **Db_xref** [defined at Identifiable]
- to one **Annotation** [defined at Identifiable]

- to one **Phenotype** [defined at Identifiable]
- to one **Identifiable** [defined at Publication]
- to one **Bibref_description** [defined at Bibliographic_reference]
- to one **Bibref_subject** [defined at Bibliographic_reference]
- to one **Bibref_scope** [defined at Bibliographic_reference]
- to one **Entry_status** [defined at Bibliographic_reference]

9 Platform Specific Model - PML

The previous chapters define a platform independent model. The real implementations, however, are expected to depend on a more specific platform. This chapter shows platform specific model, Polymorphism Markup Language (PML). Polymorphism Markup Language is a XML-based platform specific model expressing exchange format for data that conforms with platform independent model described above.

The Polymorphism Markup Language 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 Polymorphism Markup Language 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/)

9.1 Creation

The XML Schema expressing this platform specific model was obtained by converting a normative XMI file into XSD file using an SNP specific tool XSDMaker. The tool is freely available, and it is also included in the accompanied file of this specification, including its documentation.

Several XML samples were manually created. They have been validated by the XML Schema created in the previous paragraph.

9.2 Example mappings

There are several databases storing SNP information. The PML, as a data exchange format, is capable to transport their data. The following examples are non-normative.

9.2.1 HapMap

This is an example how to map PML and HapMap[1].

The tables below describe how the key XML elements/attributes correspond between PML and HapMap XML. The first table shows the correspondences of HapMap XML elements with PML. When a HapMap XML element maps to a PML attribute, it is shown using the *<element name>.<attribute name>* notation.

HapMap XML	PML
Genotype_set	Panel
Genotype	Assayed_genomic_genotype
Status	Assayed_genomic_genotype.assay_failure
Contact	Contact

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[1] Prepared by Albert Vernon Smith, Cold Spring Harbor Laboratory. HapMap details can be found at http://www.hapmap.org/.

SNP	Genomic_polymorphism
Sequence	Reference_genomic_landmark
Variation	Genomic_polymorphism
Flank_3	Genomic_polymorphism.flank_down
Flank_5	Genomic_polymorphism.flank_up
Genomic_location	Reference_genomic_location
Assembly_version	Reference_genomic_assembly
Coordinates	Reference_genomic_location
Neighbour_snp	Neighbour_variation
Assay	Polymorphism_assay
Seq_for_oligo_design	Oligo
PCR_primer_forward	Oligo
Extension_probe	Oligo

The second table shows the correspondences of HapMap XML attributes with PML (attributes are identified using the *<element name>.<attribute name>* notation).

HapMap XML	**PML**
Genotype.allele1	Genomic_allele.sequence
Genotype.allele2	Genomic_allele.sequence
Genotype.sample	Individual
Genotype_set.assay	Polymorphism_assay
Genotype_set.panel	Panel
Coordinates.chrom	Reference_genomic_location.chromosome_name
Coordinates.start	Reference_genomic_location.start
Coordinates.stop	Reference_genomic_location.end
Coordinates.strand	Reference_genomic_location.strand
Allele.base	Genomic_allele.sequence

The translation between HapMap XML and PML can be implemented easily by using XSLT.

9.2.2 ALFRED

This is an example how to map PML and ALFRED/XML[2].

The tables below describe how the key XML elements/attributes correspond between PML and ALFRED/XML. The first table shows the correspondences of elements between ALFRED/XML and PML.

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[2] Prepared by Kei-Hoi Cheung, Yale University. ALFRED details can be found at http://alfred.med.yale.edu/alfred/.

ALFRED/XML	**PML**
Population	Population
Sample	Panel
Locus	Reference_genomic_landmark
Site	Genomic_polymorphism
Typing_method	Polymorphism_assay
Allele	Genomic_allele
Frequency	Genomic_allele_population_frequency
Publication	Publication
Contributor	Contributor

The following table shows the correspondences of attributes between ALFRED and PML (each attribute is identified using the <*element name*>.<*attribute name*> notation).

ALFRED/XML	**PML**
Population.name	Population.name
Population.description	Population.ethnicity
Population.min_latitude	Geographic_location.min_latitude
Population.max_latitude	Geographic_location.max_latitude
Population.min_longitude	Geographic_location.min_longitude
Population.min_longitude	Geographic_location.min_longitude
Sample.sample_size	Panel.size
Sample.name	Panel.name
Locus.name	Reference_genomic_landmark.name
Locus.chromosome	Reference_genomic_location_in_assembly.chromosome_name
Locus.band_position	Cytogenetic_location.name
Site.name	Genomic_polymorphism.name
Typing_method.name	Polymophism_assay.description
Allele.name	Genomic_allele.name
Frequency.allele_frequency	Genomic_allele_population_frequency.value
Contributor.name	Contributor.name
Publication.title	Publication.title
Publication.date	Publication.creation_date
Publication.authorlist	Publication.authors

The translation between ALFRED/XML and PML can be implemented easily by using XSLT.

9.2.3 dbSNP

This is a comment how dbSNP can accept genotyping information in PML and redistribute it again in PML[3].

The dbSNP schema was developed before reference genomes were available, so the class concept of a sequence variation was developed around the assay process itself. The database modeled the process of polymorphism discovery as a set of submitter-driven submissions. Submissions associate the contributor with a set of accessioned assay results and associations to method objects (lab protocol or software package), population objects (a DNA sample or DNA sequence), and publication citations that describe the experiment. Each result associates identifiers of varying scope with a set of observed alleles and surrounding DNA sequence.

dbSNP also supports submissions for allele frequency, genotype frequency, and individual genotypes. While frequency/genotype data are linked to assays and organized by submitter, they are not issued accessions in the database as a distinct result. In the near future dbSNP will support submissions for haplotypes. Haplotypes, like assay results, are modeled as hierarchical sets of entities for alleles, map locations, methods, and frequencies and dbSNP anticipates the need to accession these objects as well.

The submission design assigns equal credit to all submissions of base content (assay) by accessioning content at the granularity of an assay result. The submission process is inefficient, however, with redundancy in submission content being generally proportional to the heterozygosity.

dbSNP creates a non-redundant set of submissions by grouping multiple results for the same polymorphism together in an aggregate RefSNP object. dbSNP processing assesses the complexity of flanking sequence, and when possible computes a set of locations and then uses these data to define a submission cluster in terms of identical mapping results. This process produces the reference_genome_location positions of the PML.

Since dbSNP accepts submissions for any organism, there is a secondary process to identify and group redundant submissions. When taxa lacks sufficient DNA sequence to reliably distinguish submissions by position in the genome, clusters are created based on flanking sequence similarity alone. At the opposite extreme, situations exist where assemblies across multiple taxa, and as we enter the era of cheap resequencing, assemblies will become individualized and the role of a common coordinate system may be diminished.

This is an important conceptual difference, as dbSNP may occasionally receive data with insufficient information to express the data as a variation in PML. To date, we estimate this to be a minimal fraction of dbSNP content. If serious problems are encountered, we would approach the PML draft committee to consider extensions to express variations when a DNA sequence is unavailable for larger context.

The redundancy of content in dbSNP appears as a distinctive feature of its repository role. There may be classes of data (e.g. individual genotypes) where submissions are distinct by association to an assay, but redundant when associated by RefSNP, which like the PML, represents a single genome location. It is unclear how to express dbSNP's redundancy of content when the data are transformed into PML.

I expect the vast majority of dbSNP data will be "representable" in the PML format. Limitations of the PML specification will undoubtably be revealed when we write the code to export the complete content of dbSNP. PML will stand as a useful addition to the multiple formats currently supported for redistribution.

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[3] Prepared by Steve Sherry, dbSNP project supervisor, NCBI/NLM/NIH, USA. dbSNP details can be found at http://www.ncbi.nih.gov/SNP/.

dbSNP's schema frequency evolves, as it must keep pace with new technology or new infrastructure resources demand schema revisions to capture the data. This evolution within our schema may create a class of data that cannot be mapped to PML. NCBI will ensure public access to complete content through FTP access to alternative formats. As technology expands content in variation databases, the PML schema must also evolve to maintain its significance as an interface language. Based on my experience with dbSNP and similar experience with the International Sequence Database Collaboration, annual efforts to propose and define extensions to the standard should be planned.

9.2.4 Chinese Population Genetic Diversity

This is a comment about the mapping between the Chinese Population Genetic Diversity database and PML[4] .

All the items in the database can be mapped into PML.

9.2.5 HGVbase

This is a comment about mapping between HGVbase and PML[5].

The HGVbase database, the Human Genome Variation Database, collects human genetic variations from publications, other databases and by direct submission and annotates them.

The database contents can be easily mapped into PML.

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[4] Prepared by Tieliu Shi, Shanghai Institutes for Biological sciences, Chinese Academy of Sciences.

[5] Prepared by Anthony Brookes, Head of the HGVbase database, Leicester University, UK. HGVbase details can be found at http://hgvbase.cgb.ki.se/.

A Accompanied Files

The part of the specification is a set of the accompanied files (document number dtc/05-06-03). 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 files take precedence.

A.1 Normative accompanied files

- **omgsnp-xmi<version>.xml** - an XMI representation of the platform independent model.
- **pml.xsd –** an XML Schema definition of the Polymorphism Markup Language (representing a platform specific model).

A.2 Non-normative accompanied files

- SNPclasses_associations.html – details of PIM expressed in an HTML format (the contents id the same as the section “Model classes, attributes and associations (details)” of this document).
- *.png - diagrams exported from the PIM, providing better resolution than the same pictures included in this document.
- sample*.xml – PML examples.
- glossary.html - a list of used terms.