

WHAT IS HOOM (THE HPO - ORDO ONTOLOGICAL MODULE)?

V1

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INTRODUCTION

Orphanet provides phenotypic annotations of the rare diseases in the Orphanet nomenclature using the Human Phenotype Ontology (HPO). HOOM is a module that qualifies the annotation between a clinical entity and phenotypic abnormalities according to a frequency and by integrating the notion of diagnostic criterion. In ORDO a clinical entity is either a group of rare disorders, a rare disorder or a subtype of disorder. The "clinical entity" branch of ORDO has been refactored as a logical import of HPO, and the HPO-ORDO phenotype disease-annotations have been provided in a series of triples following the OBAN model in which associations, frequency and provenance are modelled. HOOM is provided as an OWL (Ontologies Web Languages) file, using OBAN model, the Orphanet Rare Disease Ontology (ORDO), and HPO ontological models. HOOM provides extra possibilities for researchers, pharmaceutical companies and others wishing to co-analyse rare and common disease phenotype associations, or re-use the integrated ontologies in genomic variants repositories or match-making tools. HOOM is dedicated to bioinformatics use.

ORDO is accessible on several websites and updated twice per year:

- [Link](#) to HOOM on Orphadata website
- [Link](#) to SPARQL Endpoints for HOOM on Orphadata website
- [Link](#) to HOOM on Bioportal
- [Link](#) to HOOM on Orphanet website

You can find more information about ORDO in the document "[What is ORDO](#)".

HOOM is maintained by the European Union-funded [EJP-RD](#) (European Joint Programme on Rare Diseases) and [ERDERA](#) projects (European Rare Disease Research Alliance).



HOOM contains classes of the OBAN module: "Association", "Provenance" and "Evidence" (Figure 1). We chose to use this module in order to promote re-use and because OBAN is already used by biomedical community.

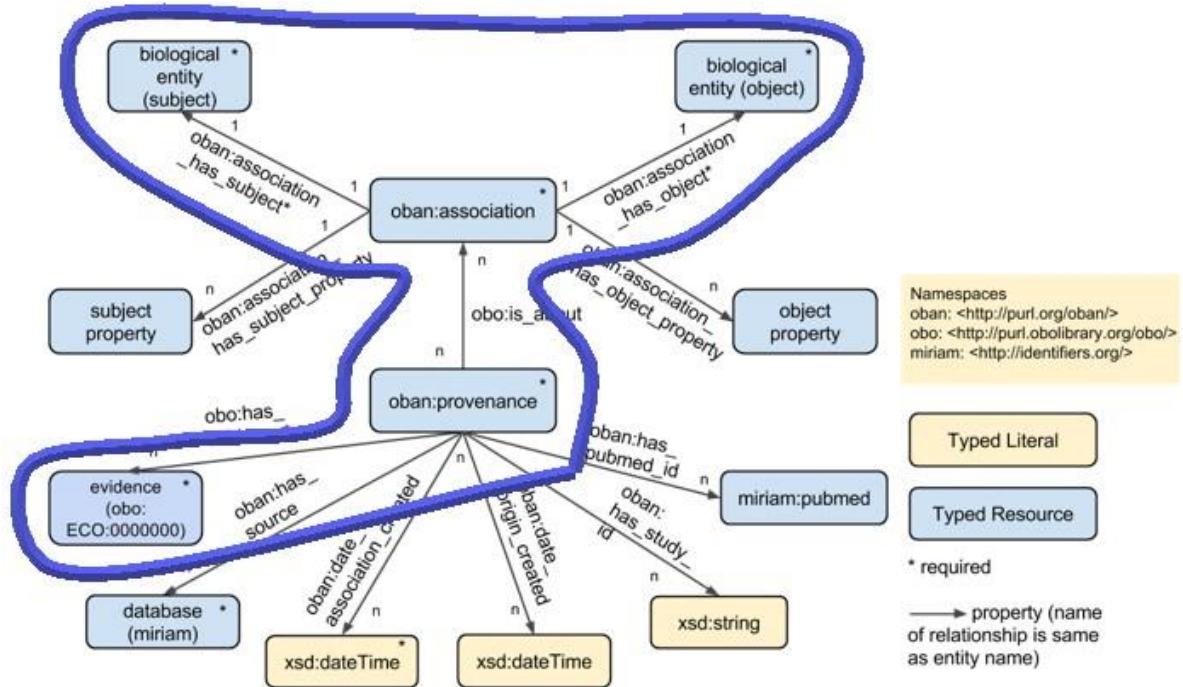


Figure 1: OBAN module. Circled in blue is the structure we reused.

The "biological entity" classes are replaced in HOOM by an "Orpha_Num" class that make the link between ORDO concepts and an "HPO_id" class that makes the link to HPO concepts. Two other classes have been created for HOOM: "FrequencyAssociation" and "DiagnosticCriteria".

Classes	Module	Label	Definition
Association	OBAN	Association	Association of a clinical entity (ORDO) with a phenotypic abnormality (HPO) according to a frequency (FrequencyAssociation). Diagnostic criterion (DiagnosticCriteria) can be attributed to the annotation.
Provenance	OBAN	Provenance	Information scientists and / or doctors of Orphanet qualifying the association (Association).
Evidence	ECO	Evidence	Scientific articles or expert advice on which the information scientist or doctor of Orphanet (Provenance) has relied to define the association (Association).
FrequencyAssociation	HOOM	FrequencyAssociation	Estimated frequency interval of a phenotypic abnormality in the patient population.



DiagnosticCriteria	HOOM	DiagnosticCriteria	The following are defined as diagnostic criteria : pathognomonic phenotypic abnormalities, phenotypic abnormalities recognised as diagnostic criteria and which have been published in scientific litterature, and the phenotypic abnormalities considered as exclusion criteria.
HPO_id	HOOM	HPO_id	Unique identifier in the HPO ontology.
Orpha_Num	HOOM	Orpha_Num	Clinical entity unique identifier in the ORDO ontology



The class "DiagnosticCriteria" has three subclasses: "criterion_DC", "exclusion_DC" and "pathognomonic_DC". The class "FrequencyAssociation" has five subclasses: "frequent", "obligated", "casual", "very common" and "very rare". For the other classes, the subclasses are considered as individuals. On schema Annex 2.b., in "Evidence" class there is "manual assertion ()" to signal that this is an assertion method that involves human review.

SubClassOf	Classes	Label	Definition
criterion_DC	DiagnosticCriteria	criterion_DC	Criterion_DC are the phenotypic abnormalities used consensually to establish the clinical diagnosis and that have been the subject of recommendations published in a peer-reviewed journal.
exclusion_DC	DiagnosticCriteria	exclusion_DC	Exclusion_DC are the phenotypic abnormalities that are still absent AND that make possible to exclude the diagnosis.
pathognomonic_DC	DiagnosticCriteria	pathognomonic_DC	Pathognomonic_DC are sufficient phenotypic abnormalities to definitively establish and undoubtedly the diagnosis.
obligate	FrequencyAssociation	obligate	The phenotypic abnormality is always present and the diagnosis cannot be confirmed if it is absent.
veryFrequent	FrequencyAssociation	veryFrequent	The phenotypic abnormality is present in 80 to 99% of cases
frequent	FrequencyAssociation	frequent	The phenotypic abnormality is present in 30 to 79% of cases
occasional	FrequencyAssociation	occasional	The phenotypic abnormality is present in 5 to 29% of cases
veryRare	FrequencyAssociation	veryRare	The phenotypic abnormality is present in 1 to 4% of cases

Properties and rules have been defined. For each property a definition has been created as well as its domain and its range (Figure 2).

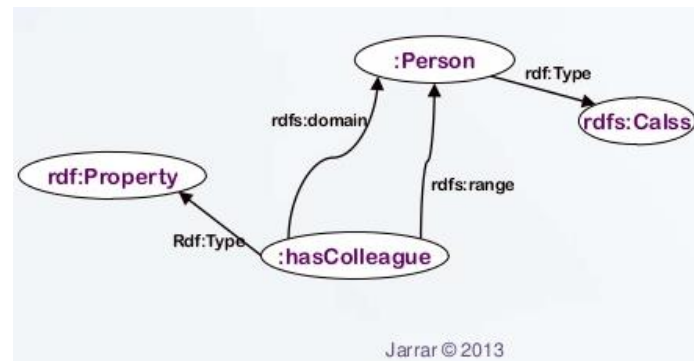
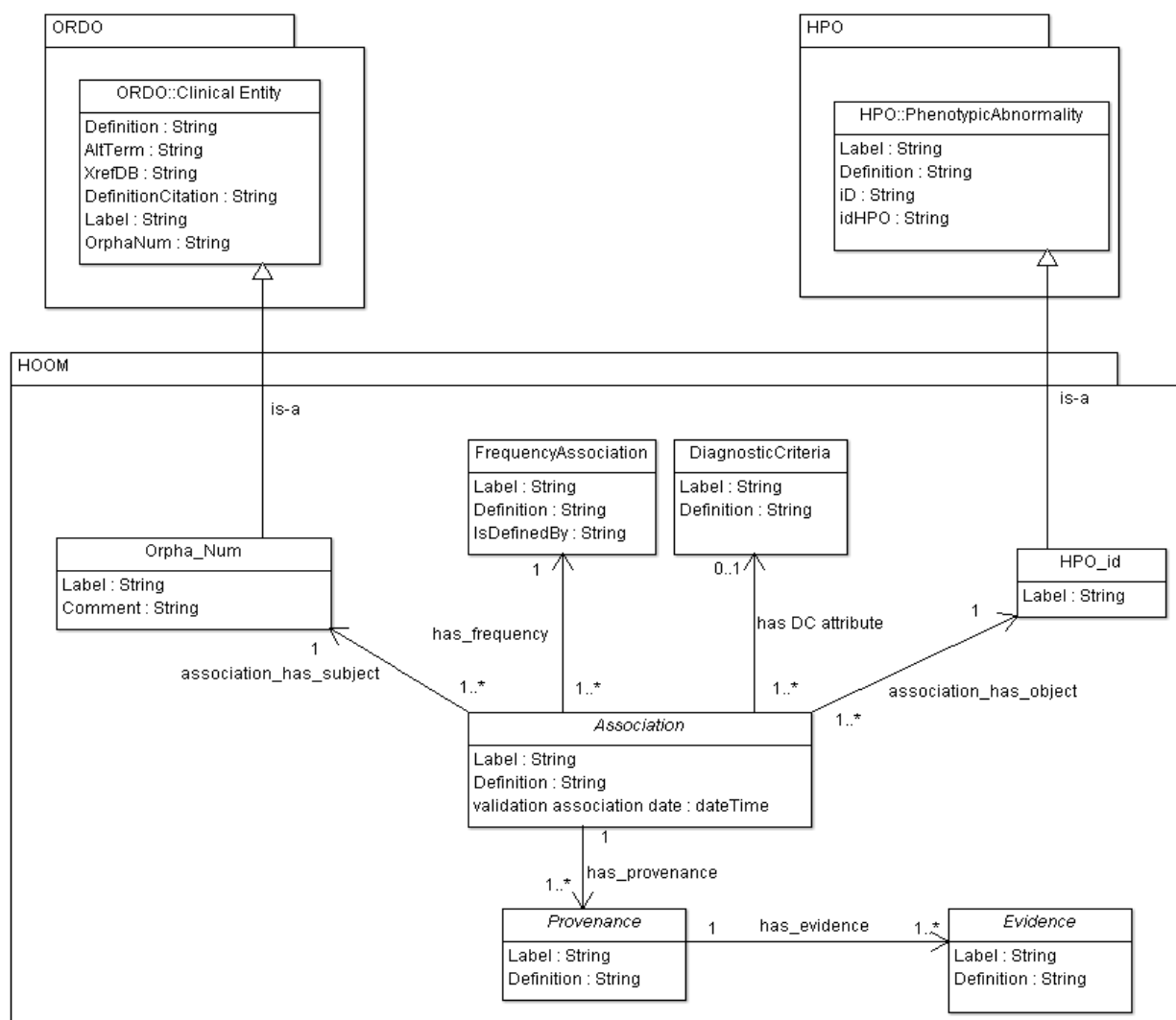


Figure 2: Example of property in RDF.

It is important to notate that because of the need to interconnect HPO and ORDO and their respective models, in HOOM there are only classes and no instances. We have made choice to have an applicative ontological module.



The HOOM Model

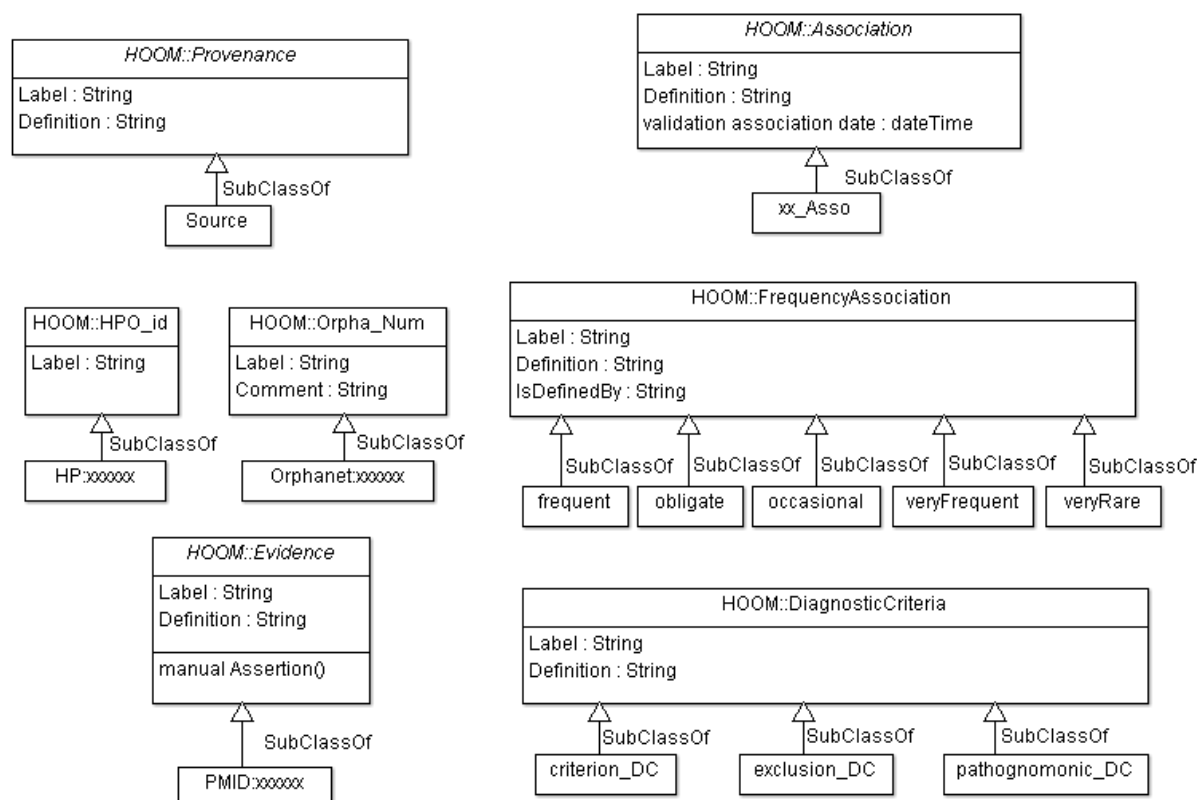


Each "association" is linked to an "Orpha_num" with a property called "association_has_subject". It is also linked to an "HPO_id" by a property named "association_has_object". A "FrequencyAssociation" is also linked by using the "has_frequency" property. These three concepts are required to create an "association".

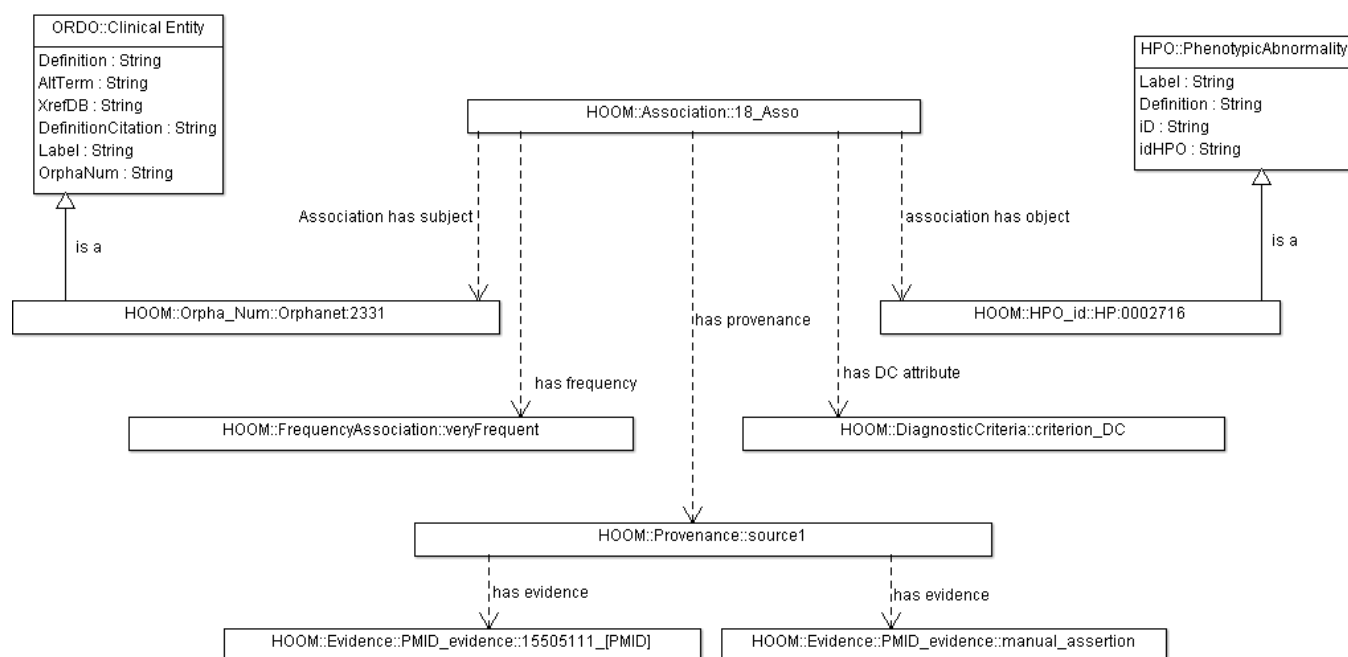
An "association" may also have a "DiagnosticCriteria" associated with the property "has_DC_attribute" and a "Provenance" associated with the property "has_provenance". Finally, "Provenance" is linked to an "Evidence" with "has_evidence" property.



Repartition of SubClassOf in HOOM.



Example of association between an ORDO Clinical Entity and HPO Phenotypic Abnormality. We used Kawasaki disease for this example.



To exploit HOOM, SPARQL queries can be use on SPARQLendpoint, virtuoso, blazegraph or other tools who permit SPARQL queries. You can use any triplestore by loading ORDO, HPO and HOOM.

These are examples of queries that you can use:

Example 1:

From an Orphanumber, getting the disease label, its associated HPO IDs with related frequencies, sources and dates of the created associations.

```
select DISTINCT ?association ?Property ?value ?date ?label ?comment
where {
    ?association owl:equivalentClass ?collection .
    ?collection owl:intersectionOf ?list .
    ?list rdf:rest*/rdf:first ?item .
    ?item owl:someValuesFrom <http://www.orpha.net/ORDO/Orphanet_2331>
    OPTIONAL{
        ?association owl:equivalentClass ?node .
        ?node owl:intersectionOf ?list .
        ?list rdf:rest*/rdf:first ?item2 .
        ?item2 owl:onProperty ?Property.
    }
    OPTIONAL {
        ?item2 owl:someValuesFrom ?value .
        OPTIONAL {?value rdfs:comment ?label} .
        OPTIONAL {?value rdfs:label ?comment}
    }
    OPTIONAL {?item2 owl:hasValue ?date .}
}
ORDER BY ?association
```

Example 2:

Obtain the number of Disorder-HPO associations.

```
SELECT (COUNT(?association) as ?Association_Amount)
WHERE {
    ?association rdfs:subClassOf ?type .
    ?association owl:equivalentClass ?node .
    ?node owl:intersectionOf ?list .
    ?list rdf:rest*/rdf:first ?item .
    ?item owl:onProperty ?Property.
    ?item owl:someValuesFrom ?Id.
}
```

Obtain the number of HPO associations for one disorder (i.e Marfan syndrome)

```
PREFIX hoom: <http://www.semanticweb.org/ontology/HOOM#>
SELECT (COUNT(?association) as ?Association_Amount)
WHERE {
    ?association rdfs:subClassOf <http://www.semanticweb.org/ontology/HOOM#Association> .
    ?association owl:equivalentClass ?node .
    ?node owl:intersectionOf ?list .
    ?list rdf:rest*/rdf:first ?item .
    ?item owl:onProperty ?Property.
    ?item owl:someValuesFrom <http://www.orpha.net/ORDO/Orphanet_558>
}
```



Authors : Orphadata Team

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<http://www.orphadata.org/cgi-bin/img/PDF/WhatIsHOOM.pdf>

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