OD4RD2 Technical Workshop Report

18 December 2024

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Content summary

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Workshop Objective

In the frame of OD4RD2 (Orphanet Data For Rare Diseases 2) we aim to ease the adoption of the Orphanet nomenclature and the implementation of ORPHAcodes in Health Information Systems by delivering the "Orphanet Nomenclature Pack" and the ORPHAcodes API (https://www.orphadata.com/orphanet-nomenclature-for-coding/) .

The main objectives of the workshop were to give an overview of the existing technical solutions and also to collect feedbacks from stakeholders already using ORPHAcodes or in the process of implementing them, in order to identify and prioritize possible technical improvements for the future releases of the tools in order to comply with the different settings and users' needs as much as possible.

Participants

Orphanet Nomenclature National hubs identified the appropriate National Institutions & technical teams, in charge of the technical implementation of coding terminologies in Health Information Systems also invitation was sent out to the JARDIN WP8 participants.

There were 109 participants of which 24 Orphanet Network members. Participants pinned themselves on the map when asked where are you from.

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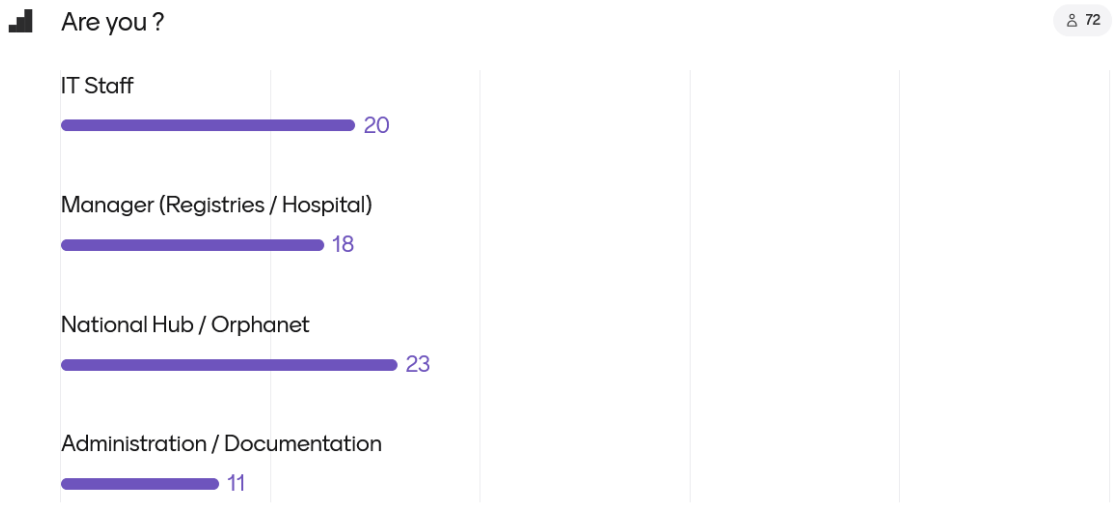
Representing the following professional categories:



Agenda

10h - 10h30 Welcome & « roundtable » https://www.menti.com/alpnsc9g76sn

Who's who (Your institution, position and ORPHAcodes implementation status : done, started, considering to start)

10h30 ORPHAcodes presentations

- Brief overview of "Nomenclature Pack" (15min)
- Where to find datasets & data access (10min)
- The HELPDESK (15min)
- Q/A session

11h20 API / apps

- ORPHAcode API (20min)
- Dataviz / Mappor / Classifications browser (20min)

12h-13h Use cases feedbacks / Discussions / Improvements

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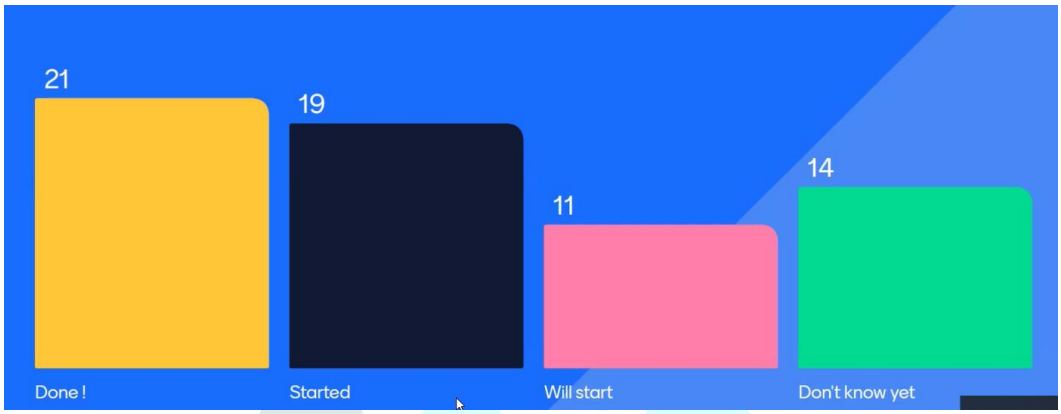
Items Presented & discussed

What is the ORPHAcodes implementation Status (participants feedback) ?



What do you need ORPHAcodes for (participants feedback)?

What is the Nomenclature Pack?

The Orphanet nomenclature pack compiles various files which provide the computable information necessary to achieve implementation of ORPHAcodes in health information systems and ensure easier and accurate coding. These files are updated every year in July to reflect the evolution of knowledge and differentials are provided to ensure traceability, as per recommendation of the RD-ACTION working group for routine maintenance of codification . It contains all the ORPHAcodes needed to code Patient RD diagnosis (i.e. Groups of disorders, disorders, subtypes of disorders & ORPHA:616874 allowing for coding yet undiagnosed patients after full investigation.

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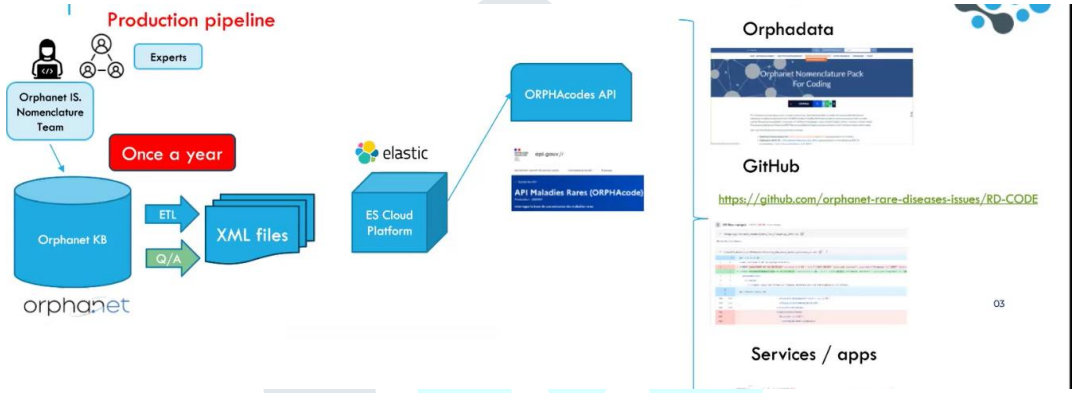
What is the Production Pipeline of the Nomenclature Pack?

Work on the Orphanet Nomenclature is carried out on daily basis by our Nomenclature Project officers, and the information is updated in the Orphanet Knowledge base weekly.

The information contained in the Nomenclature Pack is derived from the knowledge base once a year and, at the same time, the tools & ORPHAcodes API are updated.

This annual cycle was a request by the RD-ACTION working group for routine maintenance of codification in order to meet the need of having updated information reflecting the evolution of knowledge on RD but also the necessity not to burden the HIS in Hospitals.



How to access the Nomenclature Pack?

The Nomenclature Pack can be accessed via the Orphadata website: https://www.orphadata.com/orphanet-nomenclature-for-coding/

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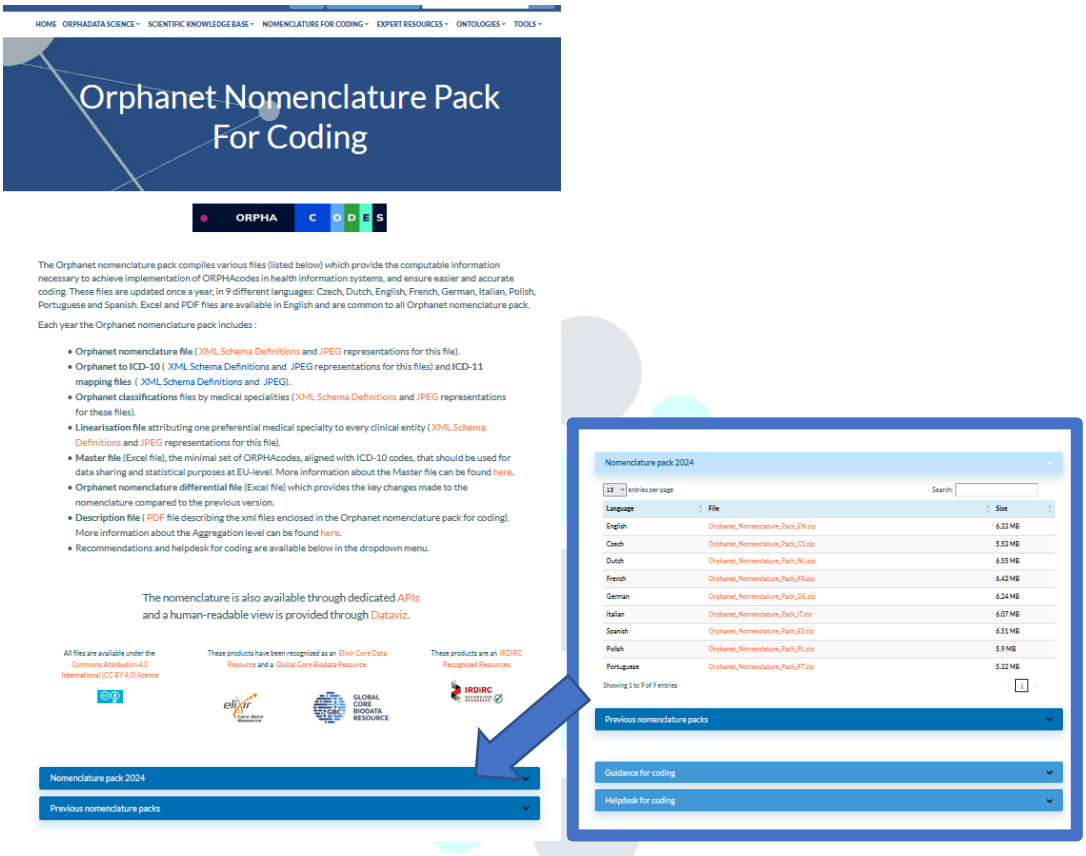
What the Nomenclature Pack contains (technical pov)?

https://www.orphadata.com/pack-nomenclature/



Nom	Taille	Compressé	Type	Modifié	CRC32
..			Dossier de fichiers		
Classifications	35 876 642	1 813 789	Dossier de fichiers	27/06/2024 14:...	
ORPHA_ICD10_mapping_en_2024.xml	9 537 420	517 671	Document XML	27/06/2024 13:...	D1A84F28
ORPHA_ICD11_mapping_en_2024.xml	6 952 962	365 856	Document XML	27/06/2024 13:...	21A1B79D
ORPHA_ICD11_mapping_en_newversion_2024.xml	8 604 815	467 269	Document XML	27/06/2024 15:...	840A62D6
ORPHAlinearisation_en_2024.xml	6 110 998	302 889	Document XML	27/06/2024 13:...	1E6F1947
ORPHAnomenclature_diff_en_2024.xlsx	64 212	54 922	Feuille de calcul Mic...	29/07/2024 16:...	1FB411AF
ORPHAnomenclature_en_2024.xml	20 004 757	1 896 340	Document XML	27/06/2024 12:...	BBBDB1C9
ORPHAnomenclature_MasterFile_en_2024.xlsx	847 216	717 286	Feuille de calcul Mic...	27/06/2024 15:...	1F4A7B1A
ORPHAnomenclaturefilescontent_2024.pdf	518 012	481 773	Document Adobe ...	04/07/2024 09:...	696914C3

The Orphanet nomenclature files for coding, or Orphanet Nomenclature Pack, consist of a set of files designed for the implementation of the Orphanet nomenclature into health information systems, that include:

- an Orphanet nomenclature file (XML) **[MAIN FILE]**
- an Orphanet to ICD-10 alignment file (XML),
- an Orphanet to ICD-11 alignment file (XML),
- a directory containing all the Orphanet classification files (XML),
- a Linearisation file (XML),
- a Master file (XLSX)
- a Differential file (XLSX)

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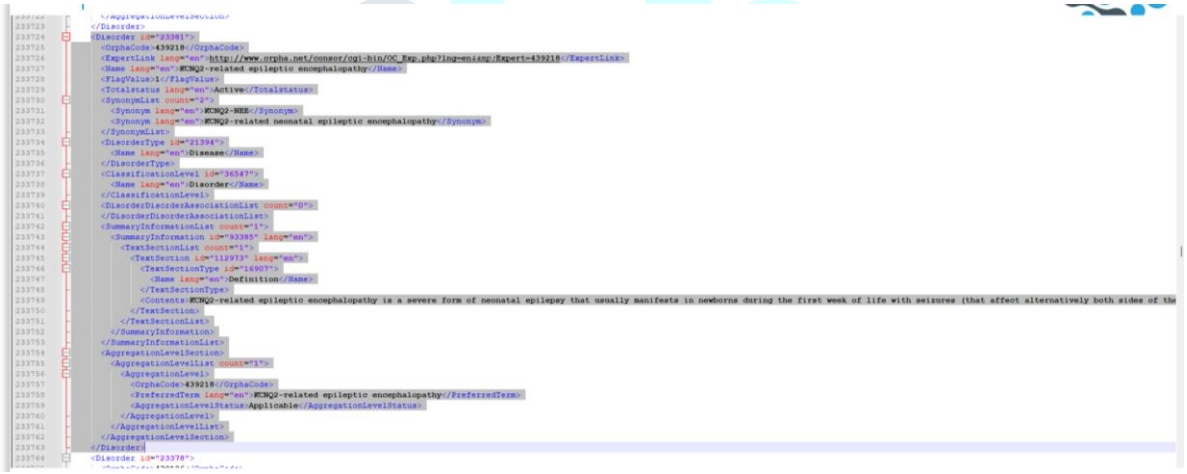
A document available in the pack & in “Guidance for coding” section describes in detail the content of these files and the way to explore them:
https://www.orphadata.com/docs/ORPHAnomenclaturexmlcontent.pdf

Why the main file and most files are delivered as XML?



Extensible Markup Language (**XML**) is a markup language and file format for storing, transmitting, and reconstructing data

It defines a set of rules for encoding documents in a format that is both human-readable and machine-readable. XML tags represent the data structure and contain metadata.

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What is the Master File contained in the Nomenclature Pack?

« MASTER » FILE

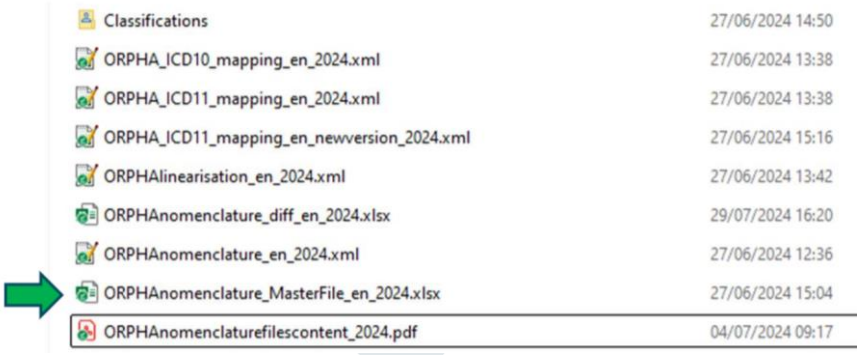
Not for direct patient coding !
The "Master File" was developed in order to support Rare Disease data sharing across Member States.

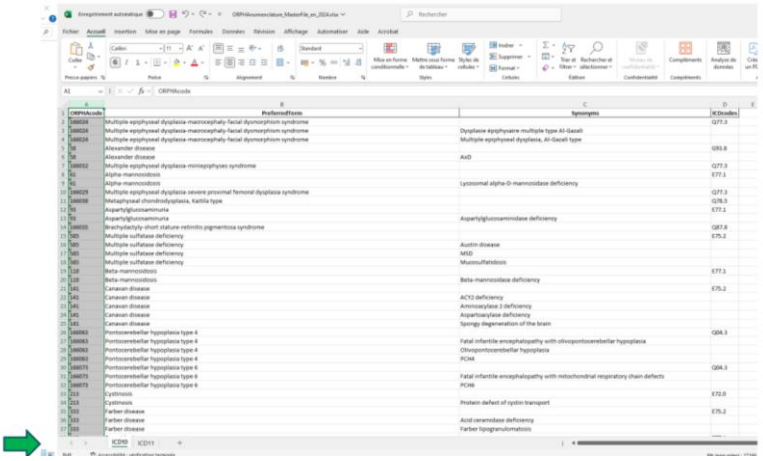
The Master File should facilitate the standardised use of the Orphanet Nomenclature by providing only the data that is relevant for data sharing at EU-level and thus enable international **statistical aggregation.**

It is a **data subset** of the Orphanet nomenclature files for coding Active ORPHAcodes registered at the Disorder level in the Orphanet nomenclature of Rare diseases
(Groups and Subtypes are excluded); **No "granularity"** for coding

Alignements available in Orphanet between these ORPHAcodes and ICD-10 / ICD-11 codes

Not part of the « API » or Apps/Services





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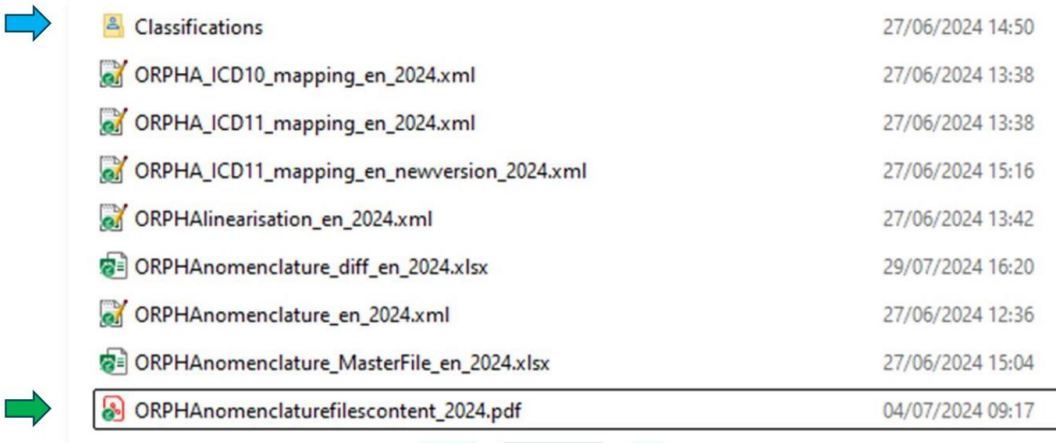
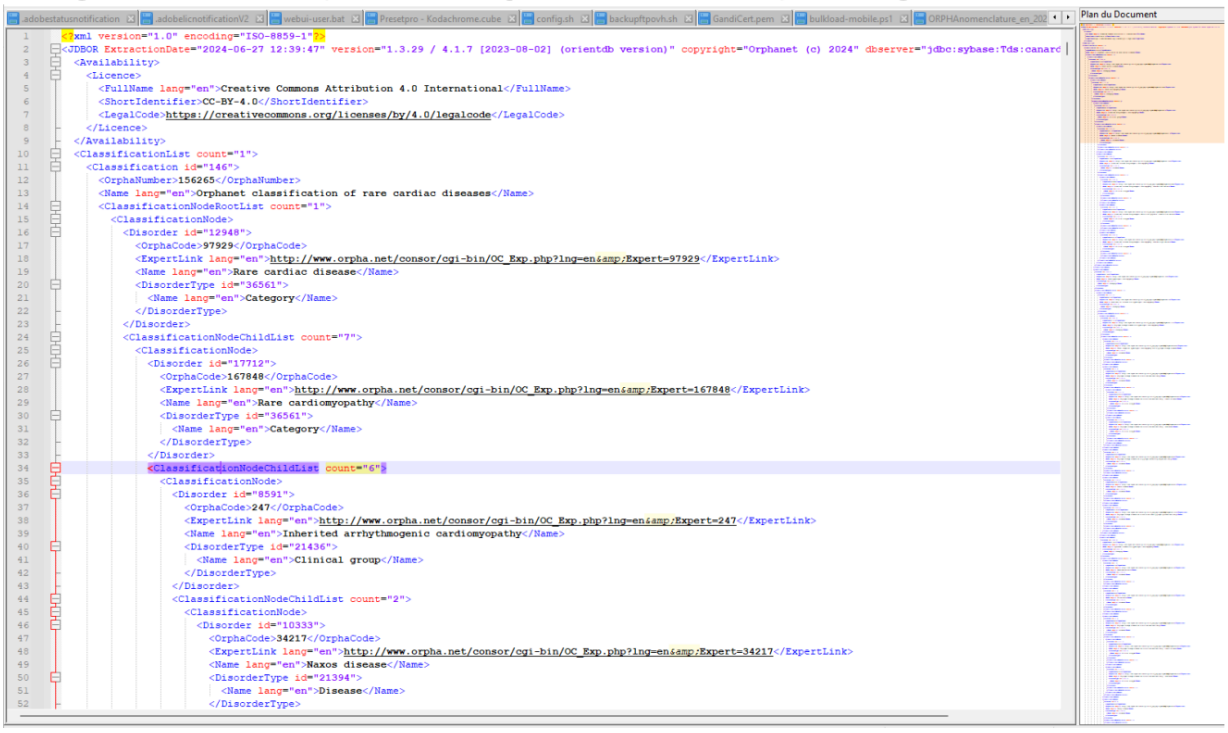
Are Orphanet Classifications Included in the Nomenclature Pack?

Yes, they are present.



As per file above:



Are previous versions of the Nomenclature Pack available?

Yes, they are available in the Nomenclature Pack files. They track changes compared to previous year release to facilitate the implementation of the Annual released information. This list of changes can help as well in comparison of data over time and to explain shifts in coded data. For a technical user this file can be an easy way to address the changes between two versions and to focus the work on primarily implementing these changes. For a full tracking of changes within the XML content, a technical user can also choose to use our dedicated GitHub repository : https://github.com/orphanet-rare-diseases-issues/RD-CODE/

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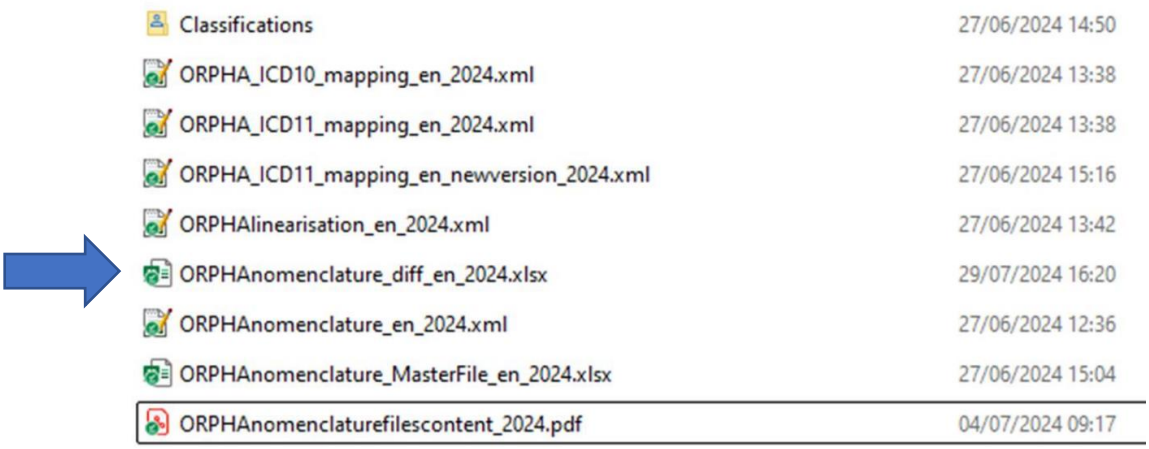
Using Github functionalities, a technical user will have a full access to every "commit", previous versions and changes.

However, a "diff" excel file is provided with each release.



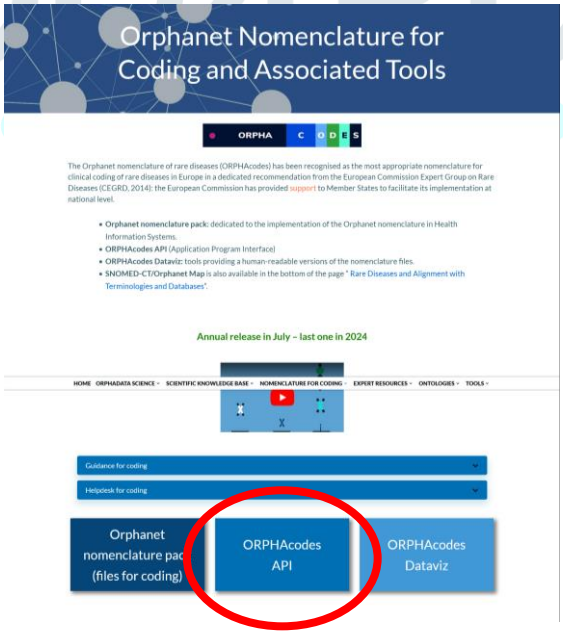
Is it difficult to include RDF export in Nomenclature Pack?

Technically doable. Also using JSON-LD. However, this could be misleading with ORDO (Orphanet Rare Diseases Ontology) which is in OWL/RDF format (see below). Nevertheless, this could be discussed for a future version if required as mandatory (not annotated as such in the whiteboard this time).

What is the ORPHAcodes API?

The ORPHAcodes API facilitates the informatic access to the Nomenclature Pack data and allows flexible implementation into the various IT systems in use in the different countries and/or settings. It also allows to build your datasets for specific national use cases.

It can be accessed from the Orphadata website: https://api.orphacode.org/



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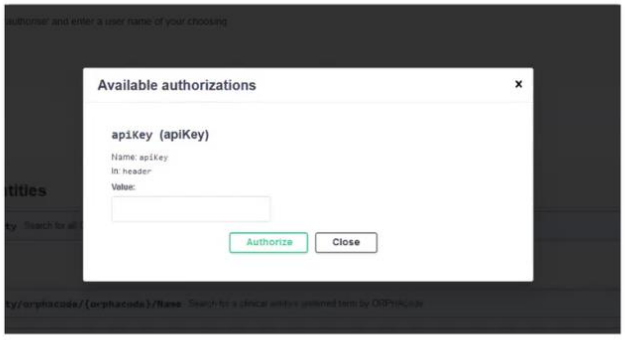
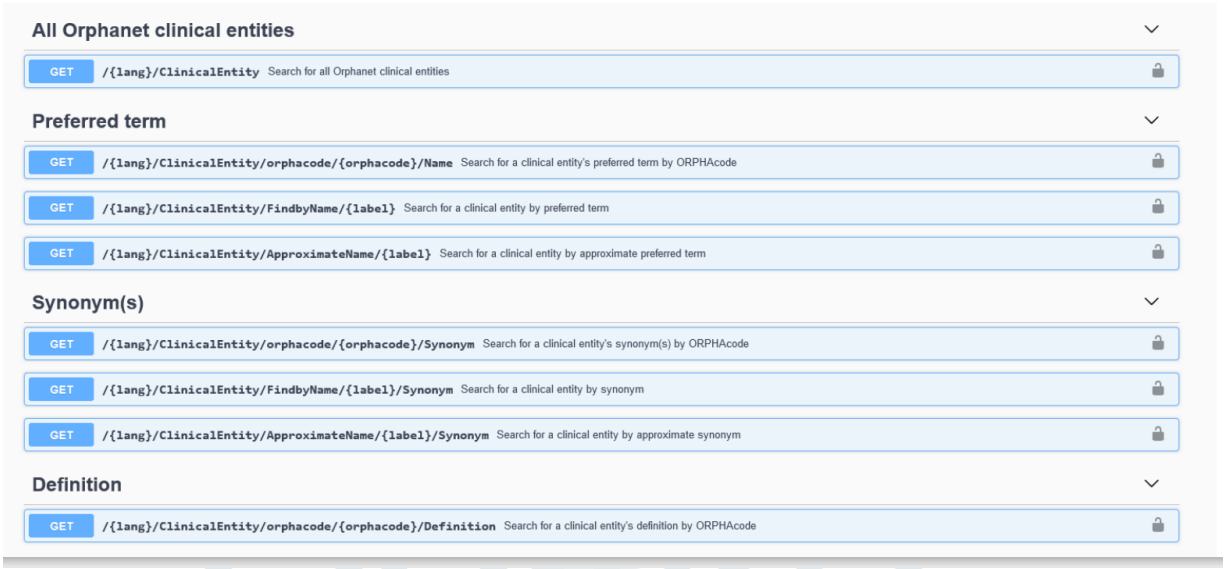
ORPHACODES API https://api.orphacode.org/

API key is needed in Header.

Nevertheless, at present time any "Key" will be considered as valid





- Orphanet provides an entire nomenclature but, locally, there could be implementation choices that are politically driven in which countries implement only a subset of ORPHAcodes depending on the national RD policy.
  This API allows to generate a sub-list of ORPHAcodes that may of interest at national level, for example only cardiac diseases or Excluding cancer and/or infectious diseases and/or intoxications => the API will provide the full list of ORPHAcodes for that particular use case.
  This adds however a layer of vigilance in terms of update at the local level as restricting the list of codes, makes the removed diseases not visible.
- The ORPHAcodes API can also be exploited for checking the validity of your code (i.e. Status: active/inactive): You can track the inactive codes, and also ask to retrieve, for deprecated entities, what is the new target code.

Take away message: The more adaptations you do locally, the more work you need to develop locally to keep up in terms of updates and follow up.

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Can I use the API front end or back end? Is there a SLA?

Please, keep in mind that there is no service level agreement (SLA) signed because we cannot guarantee you 24/7 access, however we set up a system that allows for it (but with no guarantee). For this reason, you best not to use it in production: we recommend to build your own back end once a year.

What are the planned improvements for the ORPHAcodes API?

Technical points:

❖« get » endpoints improvements? (to be discussed)

- Aggregate more data at once (less granularity which leads to use several queries “Get” instead): ORPHAcode+label+synonyms+definition+typology+status+level+url in one query.

❖ classifications, add a « get » to obtain the full list of possible classifications and classifications Ids

Important : in the Next version of the API we will add new gets but not change the previous ones which will ensure full compatibility with previous API already existing “gets”.

Is it necessary to update the ORPHAcodes API daily?

No point as its content is updated yearly in July

What are the links with ORDO/RDF?

The Orphanet Rare Disease ontology (ORDO) was initially jointly developed by Orphanet and the EBI to provide a structured vocabulary for rare diseases capturing relationships between diseases, genes and other relevant features which will form a useful resource for the computational analysis of rare diseases. The ontology is also produced according to the same pipeline as of the Nomenclature Pack: it is an export of the knowledge base of the disease content in a format that is semantically valid (links between different semantically defined concepts). However, ORDO is updated twice a year, in January and in July: only the July version corresponds to the reference Nomenclature for coding to be implemented in hospitals.

ORDO/RDF is more expressive, but the the ORPHAcodes content is exactly the same only in the July release and more updated in the following January release.

N.B. The canonical reference for coding is in the Nomenclature Pack, If you decide to use ORDO for implementation of ORPHAcodes in the HIS it is possible even if it is not the recommended way. If this is the case the July issue of ORDO should be used as this will have the same disease content of the Annual release of the Nomenclature Pack and this will allow comparability.

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What the different tools available contain? How do they differ?

As suggested a more visual representation of the content of each tool is provided in the table below.

	ORPHA codes	Classification	Master file	Differential file	Linearisation file	Genes	Genes/ diseases relation	ICD 10-ICD11	Other alignments (see table below)	Annual release	Released twice a year
Nomenclature Pack	v	v	v	v	v			v		v	
ORPHAcodes API	v	v						v		v	
ORDO	v	v				v	v	v	v		v

Table. Comparison of the of the different Nomenclature Pack & Nomenclature tools

What is the best format to use: Nomenclature pack, API…?

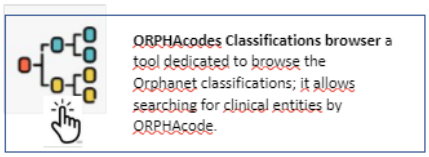
To avoid any discrepancies, for patients coding purposes, the usage of the Nomenclature Pack is recommended ("XML files"), nevertheless it is possible to use the ORPHAcodes API. Indeed, the content between "xml" files or ORPHAcodes API (json based) is fully consistent so you can choose the content that suits your system and needs the more.

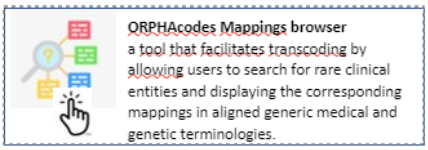
If you need to use "semantic technologies" (ontologies and reasoning, sparql, RDF/OWL etc.) you can use ORDO (with some attention with using the JULY release version of ORDO so that the content is equivalent with the July Nomenclature Pack release).

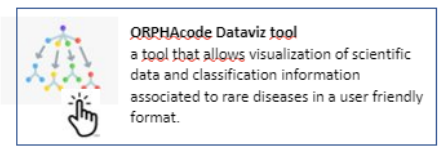
If you need to expand your datasets and include HPO annotations, genes etc.: you can consider to use Orphadata knowledge files for that. Here again, the Orphanet Knowledge base (OrphadataScience), is updated twice a year, in January and in July, therefore we recommend to use the July release files.

Are there other tools available?

Additional Human readable tools are available derived from the API (and as such updated with the July release of the Nom Pack)







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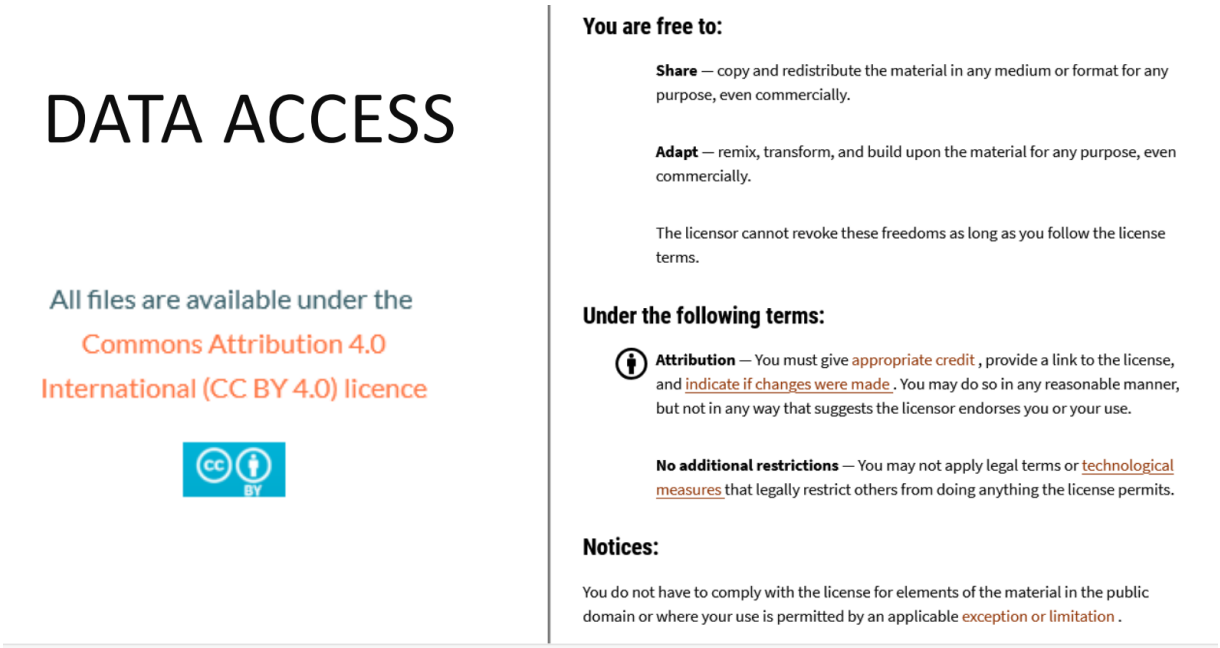
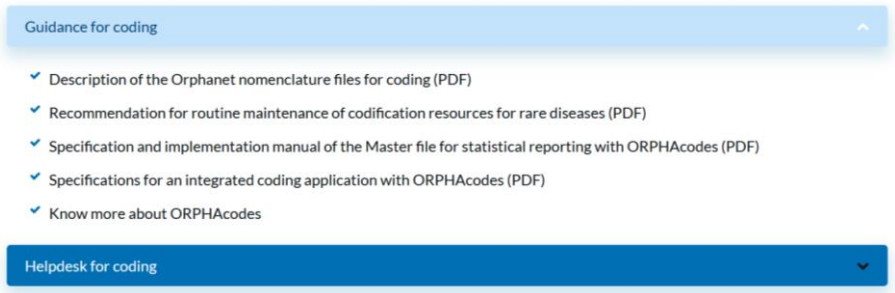
What are the conditions of data access?



Is Guidance for ORPHAcoding implementation and exploitation available?

Yes , in Orphadata we provide a PDF describing the Nomenclature Pack but also the guidelines and recommendations for ORPHAcodes implementation and exploitation released(produced during the RD-ACTION EU Joint Action and updated by the RD-CODE project) in order to ensure internationally standardised data collection.



Is a helpdesk available?

Our ORPHAcodes implementation support helpdesk has been set up in the frame of the OD4RD project. It is dedicated to answering questions related to the Orphanet nomenclature content and the implementation of ORPHAcodes in Health Information Systems or other Systems: It is an online ticketing system that allows requests to be stored, tracked and made available to others. This system facilitates an agile and interactive workflow.To post issues, please first create an account in github : https://github.com/OD4RD/Main-Help-Desk

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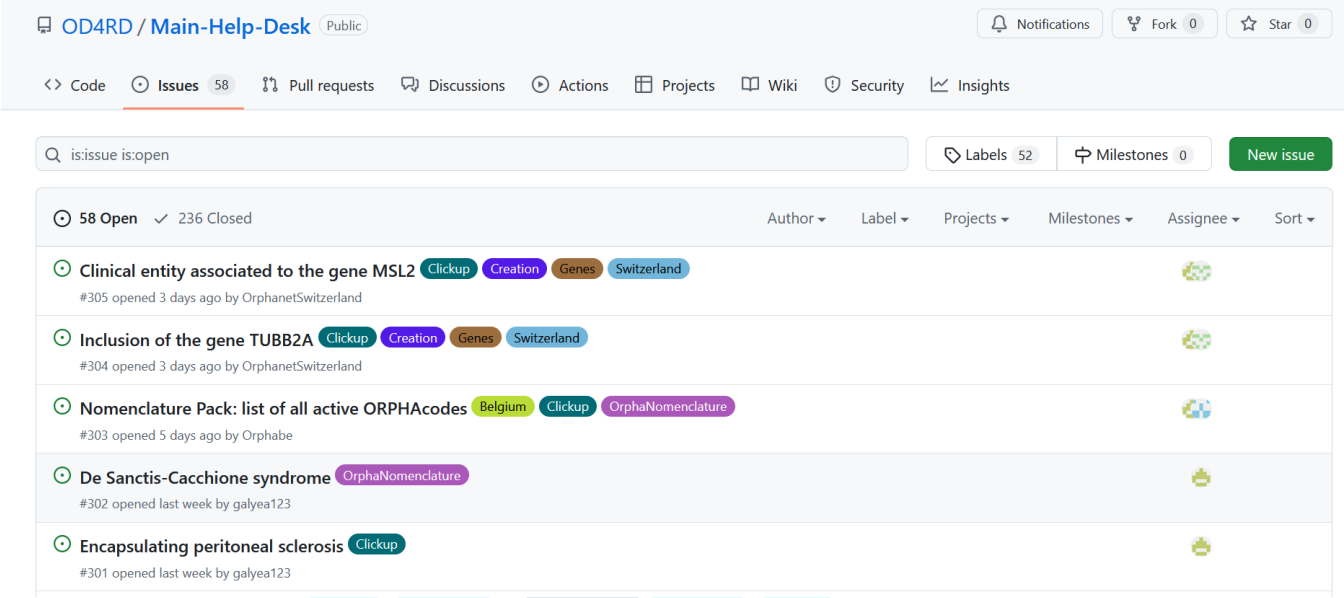
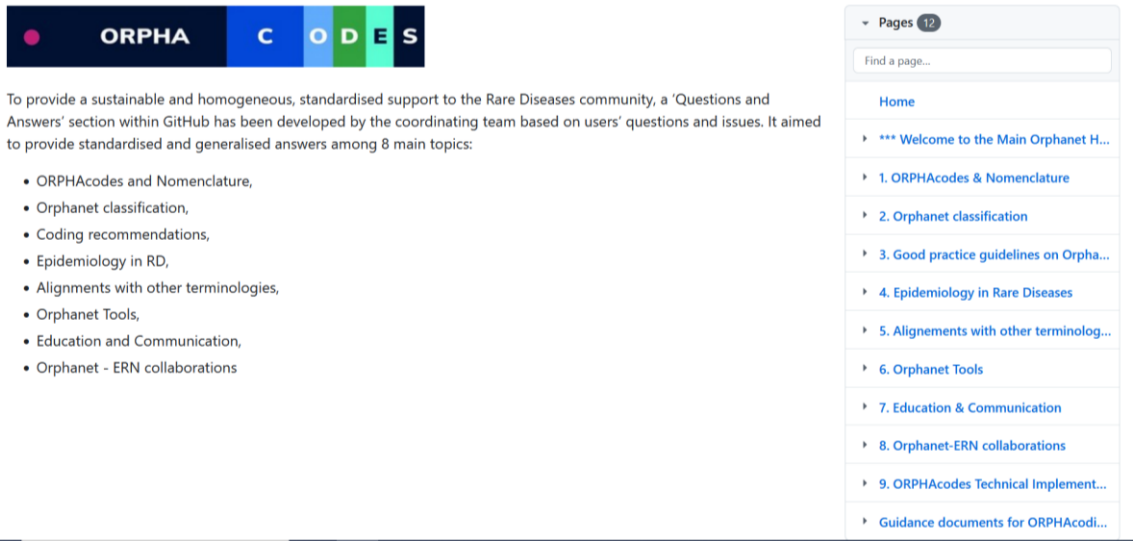
National Heldpdesks are also available and can be contacted in National Language. A new page on the Github with links to the National Helpdesk will be added to facilitate access.



To provide a sustainable and homogeneous, standardised support, a 'Questions and Answers' section within the GitHub has been developed by the Orphanet based on users' questions. It provides standardised and generalised answers among 8 main topics: alignments with other terminologies, coding recommendations, education and communication, epidemiology of RD, ORPHAcodes and nomenclature, Orphanet classification and Orphanet tools, a techy wiki . It provides also updates on the Orphanet Nomenclature-Classification revision by ERN collaborations status and links to the Guidelines and recommendation documents.

For standardized reference Orphanet answers please check the FAQ page https://github.com/OD4RD/Main-Help-Desk/wiki It is open access to any end users.



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Is a guideline for the Undiagnosed code use available?

The document Existing experiences and Guidelines about the coding of undiagnosed rare diseases patients has been issued as a consensus document on codification of suspected/undiagnosed rare diseases by the RD-CODE project in December 2021 & a Video has been released guidelines for coding undiagnosed patients

How inactivated codes are handled?

=> from a Electronic Health Record point of view:

- we have several examples of how inactivation of codes is handled the information is available here in table 2 'inactivated codes column' https://od4rd.eu/communication-material/OD4RD2_Use-Cases_OC-Implementation-DE-FR-NL-NO_2024.pdf N.B. This document will be replaced by June 2025 with an update and will be available here: https://od4rd.eu/communication-material

=> From the technical point of view

- The ORPHAcodes API can also be exploited for checking the validity of your code (i.e. Status: active/inactive) : You can track the inactive codes, and also ask to retrieve for the deprecated entities what is the new target code.
- "Differentials" file within the Nomenclature can be also checked and a technical user can also track changes within files content using GitHub (https://github.com/orphanet-rare-diseases-issues/RD-CODE)

What are all actively maintained mappings to ORPHAcodes?

Alignments between generic terminologies & ORPHAcodes enable interoperability helping to standardise the coding of rare diseases in all HIS, facilitating the identification of rare disease patients and allowing for a refined epidemiological surveillance of rare diseases thanks to improved data retrieval and analyses. In Orphanet, clinical entities (groups of disorders, disorders and subtypes of disorders) are mapped to ICD10, ICD11, SNOMED-CT, OMIM, MedDRA, MeSH, UMLS, GARD & MONDO. However not all the mapping files are available in the different tools, please refer to the table below for an overview of the available mappings in the different tool.

	ICD10	ICD11	SNOMED CT	OMIM	MEDRA	UMLS	GARD	MONDO	MeSH
Nom Pack	v	v	Not allowed*						
ORPHAcodes API	v	v							
Dataviz	v	v	Soon						
Mapping browser	v	v	Soon						
ORDO	v	v		v	v	v	v	v	v
Orphadata API	v	v		v	v	v	v	v	v

Table. Comparison of the alignments information present in the different Nomenclature Pack and Orphanet tools available. * snomed requires a per fee licence

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A human readable SNOMED CT/ORPHAcodes mapping file is released every October: see below

N.B. No mapping to LOINC as it is not a disease terminology

Standardised way in sharing mappings : not implemented yet. We need to think about that (which implies to consider such standards like SSSOM https://mapping-commons.github.io/sssom). But won't be a lightweight way to provide mappings and external references.

Can we use Transcoding files to transcode from our main generic terminology to ORPHAcodes or to ORPHAcodes to generic terminology?

The recommended approach is to implement the ORPHAcodes as an aside coding system so that the clinicians can use the specific terminology to accurately code RD diagnosis; by doing so it is possible to transcode to the suggested mapped code in the generic terminology in an automated way (and meeting also other purposes such as billing…).

If the mapping files are used to transcode from the Generic terminology to ORPHAcodes, this renders the transcoding exercise more burdensome & time consuming because not every mapping is 1:1, ORPHAcodes being more granular most of the time, according to the target terminology there can be NTB (narrower-to-broader) and BTN (broader-to-narrower).

ex. if you use for example the ICD10 code and then transcode to ORPHAcodes, it is possible thanks to the Nomenclature Pack files BUT you will have to assess the mapping because one ICD10 code can correspond to the Orphanet group of diseases rather than the granular disease (and therefore to many ORPHAcodes at the disorder level), and you will have to manually assess which is the best one to choose.

What about SNOMED CT mappings?

A SNOMED CT-Orphanet nomenclature map has been produced in XLSX format in English. It is the product of a joint project carried out under a collaboration agreement between the Institut national de la santé et de la recherche médicale (INSERM) and SNOMED International.

Based on an agreed priority set, new concepts for rare diseases as defined in Orphanet have been added to SNOMED CT and a map from SNOMED CT to the Orphanet Nomenclature of Rare Diseases has been created.

Access to the human readable map is made available from Orphadata (https://www.orphadata.com/alignments/), and the RF2 version is available from SNOMED International for Members and Affiliates from the organization's Member Licensing and Distribution Service (https://mlds.ihtsdotools.org).

It is released annually in October: be careful to ensure you are working with the latest version to avoid inconsistencies.

All these alignments concern only exact matches. In other words, one ORPHAcode is aligned to one SNOMED CT ID only if both concepts represent the same entity with the same perimeter. If not, each

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nomenclature team will review this entity according to their own procedures and decide if this mapping is kept or excluded. Because of the different inclusion criteria, a 100% equivalence between the two nomenclatures is not possible to achieve. Therefore, despite the high level of alignment between both terminologies that consents an optimal level of interoperability, the entirety of rare disease is not represented in SNOMED CT. Moreover, by definition, there is always a delay between the creation of a new ORPHAcode and the mapping with a SNOMED CT ID due to a reviewing process and the production of a definition (that is necessary to create/align a SNOMED ID). We cover the aggregation level, i.e the disorder level, (like for the MF) as it is where the interoperability level is, but for groups or subtypes it should be left to comply to its own procedures.

N.B. SNOMED CT is not a terminology dedicated to code rare diseases and it does not include any mention to specify if a given disease is rare or not.

=> Rare diseases included in SNOMED CT do not have a dedicated classification of rare diseases for data aggregation but are mixed together with non-rare diseases. Therefore, using only SNOMED CT to code rare patients will not allow to retrieve aggregated specific rare disease data nor to make any specific statistical analysis on rare diseases.

Interoperability with other systems that do not use SNOMED CT and registers of rare diseases (using ORPHAcodes) is not granted just by using SNOMED CT codification.

A question asked by the audience: It is possible to solve the entities not present in SNOMED CT with NTB or BTN matches?

- ⇨ Theoretically yes, but this is not foreseen in the current agreement. Much more effort is needed for attribution of codes to broader terms, and not sure if SNOMED can manage these kinds of mappings.

Possible to have ORPHA as a standardized vocabulary in OMOP?

Technically is doable as OMOP is supposed to have a space for standardised vocabulary We are indeed studying on how we can efficiently do this.

Are ORPHAcodes annotated with HPO terms?

Orphanet database of RD is annotated with the Human Phenotype Ontology (HPO) terms, a standardized and controlled terminology covering phenotypic abnormalities in human diseases. This work is a continuous work, aiming to cover the totality of the Ortphanet disorder level. (I)At the time of writing this report, 3914 active disorders are annotated with HPO terms, representing the 60,7% of the disorder level, and this information is available in Orphadata files (https://www.orphadata.com/phenotypes/) and in the HOOM ontology (https://www.orphadata.com/hoom/).

What about genes?

To better describe rare disorders of genetic origin, Orphanet provides information on every gene related to a rare disorder. The type of relationship between genes (causative, susceptibility, etc) and their related rare disorders is provided.

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This information includes the genetic international nomenclature, the gene typology (gene with protein product, locus, non-coding RNA), the chromosomal location, the cross-mappings with international genetic databases.

You can access aggregated datasets from Orphanet concerning genes associated with rare diseases via Orphadata: https://www.orphadata.com/genes/ and ORDO (https://www.orphadata.com/ordo/)

What about OMIM -inheritance by genes?

Orphanet applies to the WHO definition of disease: a clinically defined RD. As such, ORPHAcodes, at the disorder level (which is the level for use for diagnosis and epidemiological purposes) represent disorders defined clinically to include ALL RD (not only the genetic ones) so that they can be recognisable by a clinician and allow a patient to enter a health care pathway dedicated to the specific RD (and after receive a Molecular explanation if relevant).

OMIM is a database, and not a coding terminology, on genetic phenotypes and genes. Not everything in OMIM is rare. Overlaps between Orphanet & OMIM exists (RD of genetic origin) but the entry point in OMIM is the gene, and there are as many entries as there are genes.

And because how the concepts are mapped and in which sense, you cannot just transcode because we are on different levels of concept. For example, retinitis pigmentosa can be diagnosed clinically and confirmed genetically but not always, there are more than 80 genes that can cause it. Because of that you have more than 80 entries in OMIM while in Orphanet there is only one code because it is only one disease (same clinical manifestations).

OMIM attaches to each gene a different inheritance (but this is not homogenous across OMIM). While Orphanet does not provide directly the inheritance mode of a given gene in a disorder; Orphanet provides qualified gene-disease relationship according to the pathogenicity of the described mutations. Thus, Orphanet genes are annotated as causative (from germline or somatic mutations), modifiers, major susceptibility factors or playing a role in the phenotype (for chromosomal anomalies). When causative mutations are of germline origin, loss or gain of protein function are documented if available(https://www.orpha.net/pdfs/orphacom/cahiers/docs/GB/Orphanet_Genes_inventory_R1_Ann_gen_EP_02.pdf). Therefore OMIM inheritance can be indirectly derived using ORDO.

Whiteboard

To empower the digital collaboration and facilitate brainstorming and capture ideas, a whiteboard session was held during the meeting, as per below. Directions provided were:

A product roadmap outlines the priorities, vision, and direction of a product over time. This template can be used to collect stakeholders needs.
3 Topics:
A) Nomenclature Pack Files (format, content...)
B) API ORPHAcodes (specify new"get"/"queries"
C) Documentation / HELP DESK
Use sticky notes to propose new features with MoSCoW method annotation (i.e. On sticky note, precise if your require your demand as: M :Must have- S : Should have- C : Could have- W : Wont have (to remove an unnecessary existing feature).
Results have been incorporated into the Q/A report.

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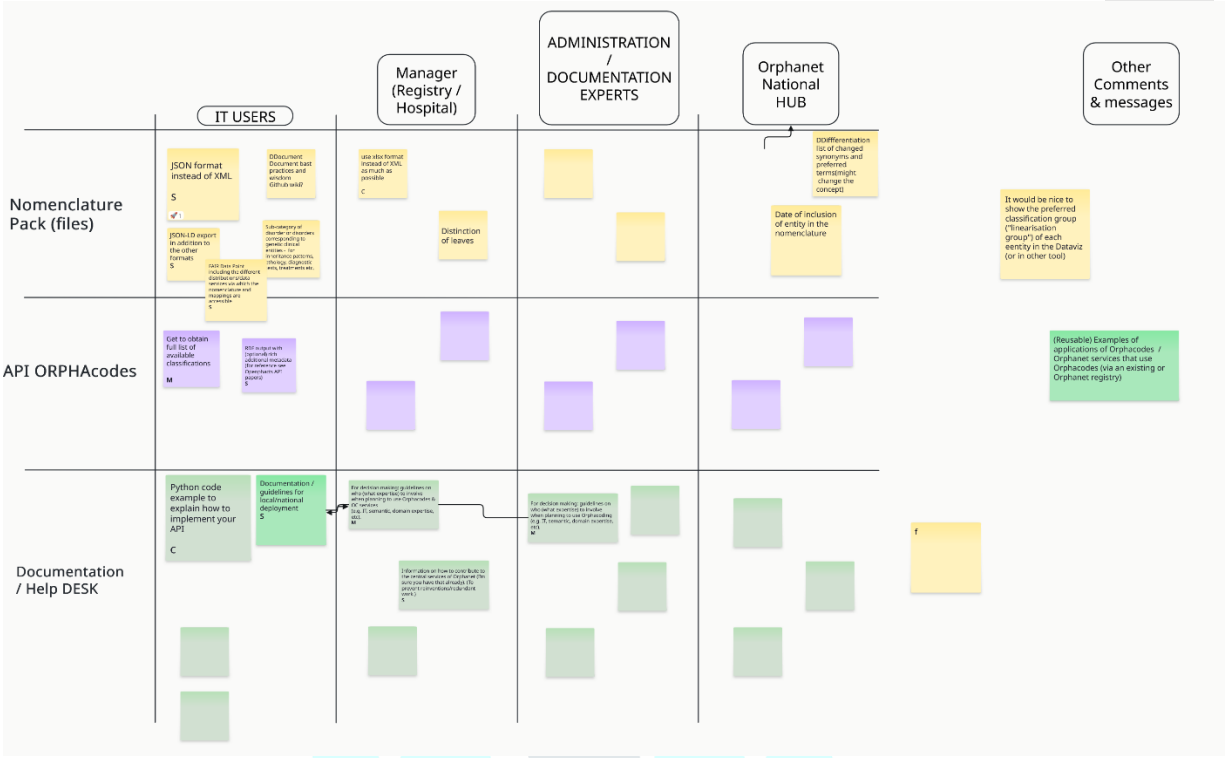

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