Data codification, standardisation, and disease Identification: ORPHAcodes and the Orphanet Nomenclature for Rare Diseases

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What is Orphanet ?

Orphanet is a unique, worldwide network, offering a complete set of tools and knowledge including a Knowledge database on rare diseases, to improve diagnosis and patient care.

CREATION
1997

42
COUNTRIES

110
PEOPLE

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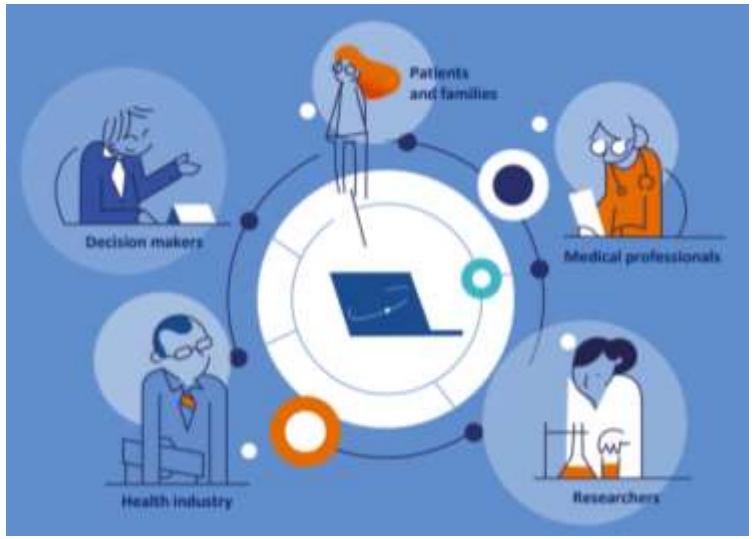


Orphanet's missions

By providing reference information and knowledge to all actors of the rare disease ecosystem, Orphanet aims to:

1

Improve the visibility of rare diseases in the fields of healthcare and research by maintaining the Orphanet rare disease nomenclature (ORPHAcodes): providing a common language to understand each other across the rare disease field.

2

Provide high-quality information on rare diseases and expertise, ensuring equal access to knowledge for all stakeholders: orientating users and actors in the field in the mass of information online.

3

Contribute to generating knowledge on rare diseases: piecing together the parts of the puzzle to better understand rare diseases.

Solving RD invisibility

Underrepresented

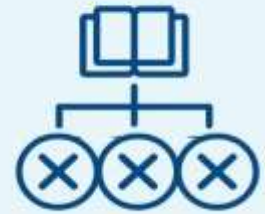
ICD-10: ~83% of RD do not have any code*
ICD-11: ~37% of RD do not have any code*
SNOMED CT: 7% of RD do not have any code*
None of these terminologies have a code for the patients in a diagnostic dead-end/undiagnosed RD patients

Imprecise

ICD-10: 93 % of RD do not have a PRECISE code*
ICD-11: 75 % of RD do not have a PRECISE code*
SNOMED CT: 7 % of RD do not have a PRECISE code*

Not classified as "rare"

RD are "lost" amongst common diseases
Generic terminologies are not exploitable for RD-specific statistics

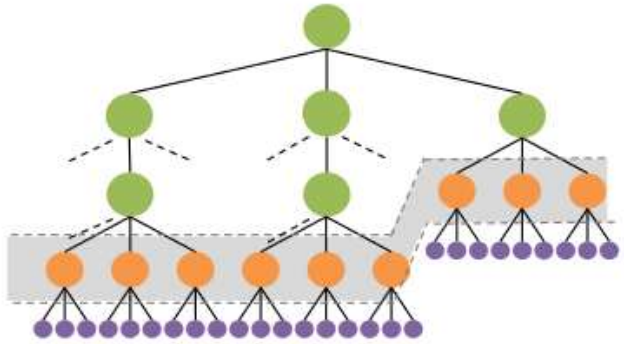


What is an ORPHAcode?

A RD-specific classification system for secondary use, including more than 6400 rare diseases

ORPHA CODES

ORPHA code	Preferred term	Synonyms	Classification level
ORPHA:2134	Atypical hemolytic uremic syndrome	Atypical HUS aHUS	Disorder
Definition	a rare thrombotic microangiopathy disorder characterised by mechanical hemolytic anemia, thrombocytopenia, and renal dysfunction.		



Group

Disorder

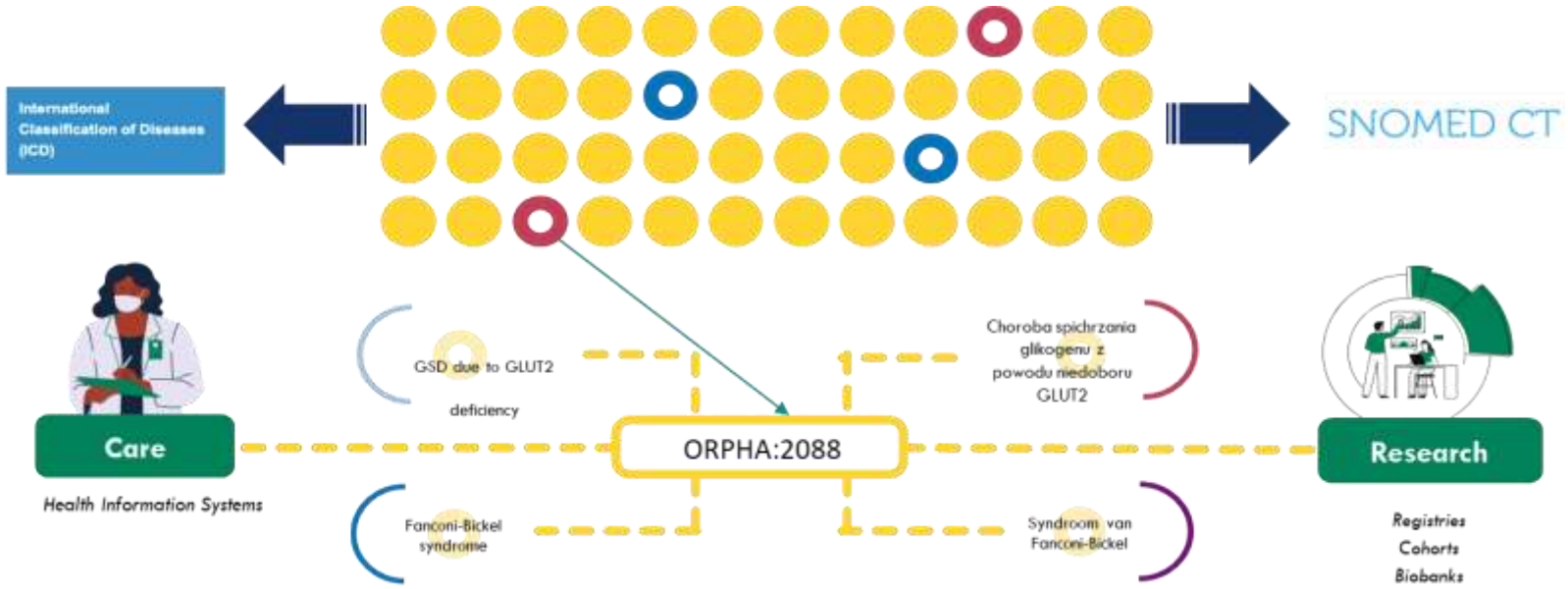
Subtype

"Classification level"

The Orphanet Nomenclature is comprehensive, standardized, evidence-based, interoperable, versioned, computable and free (CC-BY 4.0)

Rare diseases interoperability

Only ORPHAcodes can precisely identify all rare diseases



- Easier follow-up
- Easier pathway healthcare analysis
- Easier epidemiological analysis

Why orphacodes?

Only ORPHAcodes can precisely identify all rare diseases

- To ensure ALL RARE DISEASES are visible in Health information System
- To allow RD data to be interoperable among hospitals, regions, and countries
- To answer a range of public health and research questions and make evidence-based decisions

https://www.rd-code.eu/why-orphacoding/

We are ready for implementation

- **2009:** Codification as priority in the Council Recommendation for RD
- **2013**: CEGRD recommendations on Minimum Data Sets for RD
- **2014:** CEGRD recommendations on Orphacodes
- **2015-2018:** RD-ACTION guidelines on codification for harmonized statistics on RD in Europe
- **JRC's CDE** for RD registration: ORPHAcodes
- **2017:** ORPHAcodes as Best practice by the EC
- **2017:** creation of ERNs: need for data generation to monitor ERN activity and added-value
- **2018-2021:** RD-CODE: pilot ORPHAcode implementation in 4 countries; renewed recommendations; code for undiagnosed patients
- **2019:** ERN registries
- **2019:** European Joint co-fund programme for RD (EJP-RD): ORPHAcodes in standardized data models for data exploitation
- **2020-2023:** X-eHealth: approved specification for ePatient summary V3 including RD for cross-border unplanned care (ORPHAcodes)
- **2022-2025:** OD4RD & OD4RD2: Network of national Orphanet nomenclature hubs in 20 countries: local support for ORPHAcodes implementation and codification
- **2023- ongoing**: EHDS Pilot2 project: Orphanet participation to data models and interoperability workpackages (after having participated in TEHDaS)
- **2024:** JARDIN JA (Orphanet leads task 8.2 on semantic harmonization for RD data in European health information systems)

ORPHAcodes National Hub

- Trainings
- Workshops
- Helpdesks

OD4RD
Orphanet Data For Rare Diseases

EURORDIS European Rare Diseases Research Alliance

orphanet KNOW THE RARE FOR BETTER CARE

Policy for codification of RDs

Improved codification for rare diseases is cited as a priority in the Council Recommendation on an action in the field of rare diseases (2009)

3 Joint Actions from 2009 to 2018 supported the development of ORPHAcodes

2015-2018: RD-Action Joint Action : Guidance for implementing ORPHAcodification

2018- 2021 RD-CODE: Update of guidelines for implementing ORPHAcodification

2021-2022: RD-Action Joint Action : Guidance for implementing ORPHAcodification

2018- 2021 RD-CODE: Update of guidelines for implementing ORPHAcodification

2022: OD4RD pilot Direct Grant: Guidance for implementing ORPHAcodification in National Hubs

2023: OD4RD2 Direct Grant: Guidance for implementing ORPHAcodification in National Hubs

RD-action guidelines for codification

RD-ACTION GUIDELINES 2015-2018: REVISION BY RD-CODE 2018-2021

www.rd-code.eu

Application domains

- HEALTH CARE PLANNING:

  HEALTHCARE organisation planning: Use ORPHACodes plus the main routine codification system + diagnosis assertion degree

- EXPERT CENTRES:

  DATSET to COLLECT: ORPHAcode + additional elements such as phenotypic traits, genomic information...

- RESEARCH:

  DATASET to collect: ORPHAcode as granular as needed + additional element such as phenotypic traits, genomic information….

- TRANS-NATIONAL STATISTICS:

  International comparability: ORPHACode statistical aggregation level

- DATA EXCHANGE FOR CROSS-BORDER CARE:

  Minimum Dataset for care: ORPHAcodes (diagnostic level) + additional elements (depending on planned or unplanned health care use case)

Rare Diseases coding guidelines

I

Several tools and strategies could be set at MS level to produce data or statistics for RD, nevertheless each country should set this strategy accordingly to a standard principle of maximasing exhaustiveness as well as possible to re-use of existing data collections

II

Code the data in a way that the reporting can compile to the granularity of the international recommended list of ORPHAcodes ('Masterfile'- granularity). If no further national needs for reporting are necessary, use the codes from the 'master file' directly

III

Whenever possible capture the information of the diagnostic assertion for all RD cases. Use the options: 'Suspected RD', 'Confirmed RD' and 'Undertermined diagnosis'. Additional options might be helpful

IV

Update your coding resource according to the internationally agreed cycle annually in order to have the most recent coding file and to ensure comparability

V

Keep track, for each patient file, of the different ORPHAcodes and associated versions that were used to describe the patient's diagnostic pathway

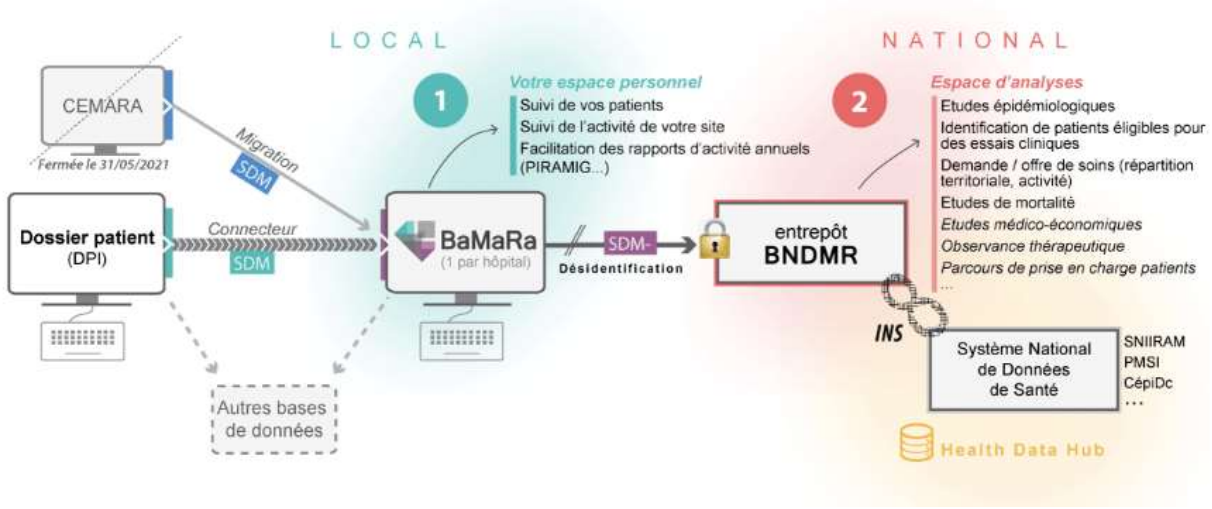
VI

If ORPHAcodes are used together with another national coding system for morbidity coding, the two systems should be linked in a standardized way to ensure that code combinations are standardized and the coding effort for the user is minimized

BNDMR (Banque Nationale de Données Maladies Rares)

Funded by the French Ministry of Health, this national database aims to provide France with a standardized collection of data based on a minimum data set. Its goal is to document the healthcare and medical status of patients with rare diseases treated in French expert centers, while also improving the evaluation of the impact of national plans on these conditions.

BNDMR
Banque Nationale de Données Maladies Rares



Guideline I

Maximise exhaustiveness & re-use, can also apply to registries

Guideline 1 - Several tools and strategies could be set at MS level to produce data or statistics for RD, nevertheless each country should set this strategy accordingly to a standard principle of maximizing exhaustiveness as well as possible re-use of existing data collections

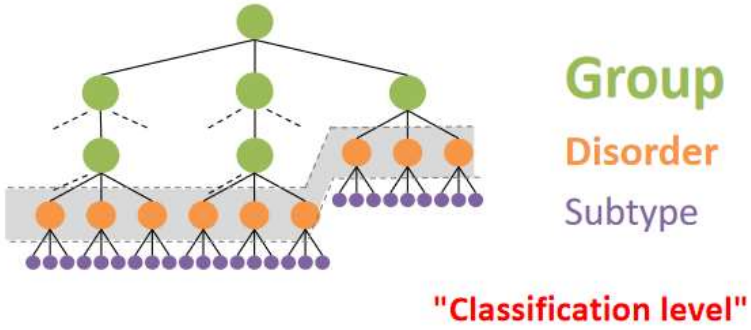
BaMaRa

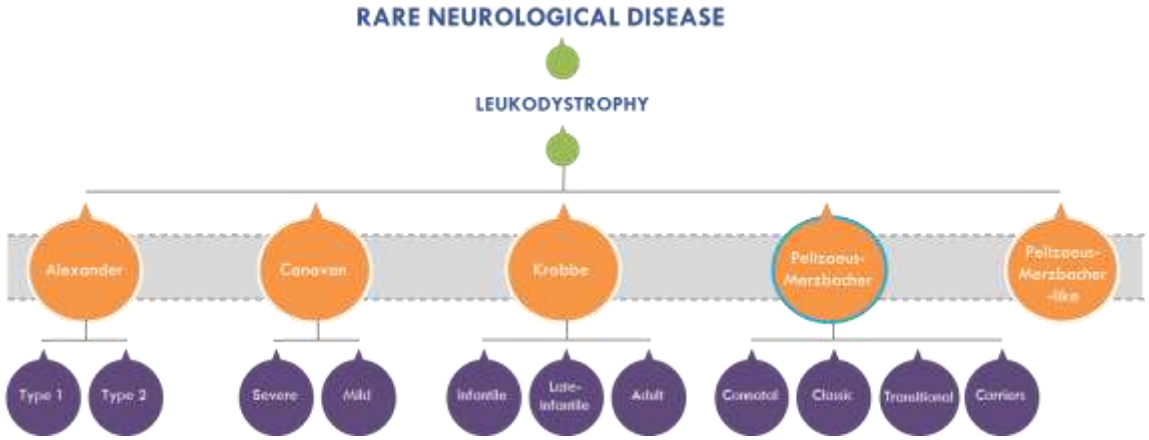
BaMaRa is an application used in French expert centers for collecting and retrieving data from the national minimum data set for rare diseases during patient care. It allows for direct data entry or interoperability with hospital electronic medical records, providing healthcare professionals with a secured and structured system to track patient care and generate activity reports.

Guideline II

Choose the good granularity level, the recommended aggregation level for statistical purposes is the DISORDER level

Guideline 2 - Code the data in a way that the reporting can compile to the granularity of the international recommended list of Orphacodes (“master file”-granularity). If no further national needs for reporting are necessary, use the codes from the “master file” directly.





Guideline III

Capture the diagnostic assertion

Guideline 3 - Whenever possible capture the information of the diagnostic assertion for all RD cases. Use the Options "Suspected rare disease", "Confirmed rare disease" and "Undetermined diagnosis". Additional options might be helpful.

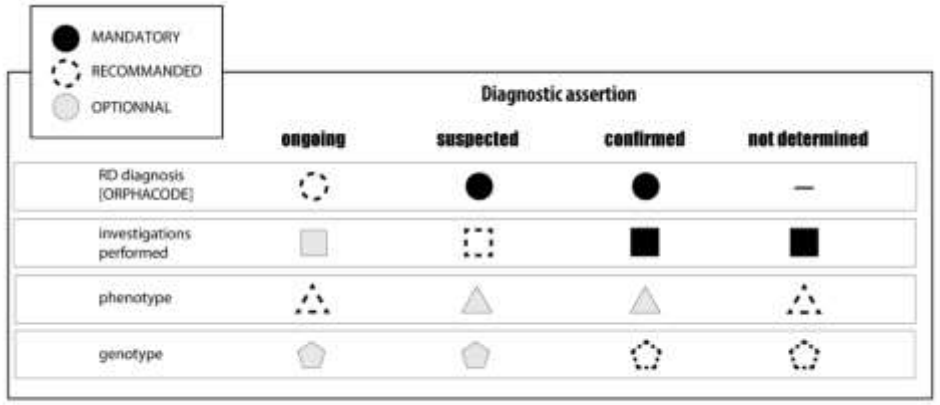


Figure 3 - RD patient description: Diagnosis, Investigations, Phenotype and Genotype

Coding undiagnosed patients

3 recommendations for coding undiagnosed patients in Electronic Health Records (EHRs) or in registries

- Capture the diagnostic assertion for all RD cases, whenever possible. However, modifying hospital EHRs or registries existing forms to collect new items might be challenging.

- Use the new code ORPHA:616874 ‘Rare disorder without a determined diagnosis after full investigation, alone or in addition to the diagnostic assertion’.

- Provide a phenotype and a genotype description of undiagnosed patients, moreover in registries.

Coding undiagnosed patients

Orphanet disease page for “Rare disorder without a determined diagnosis after full investigation”

Guideline VI

Standardized interoperability

Guideline 6 - If Orphacodes are used together with another national coding system for morbidity coding, the two systems should be linked in a standardized way to ensure that code combinations are standardized and the coding effort for the user is minimized.

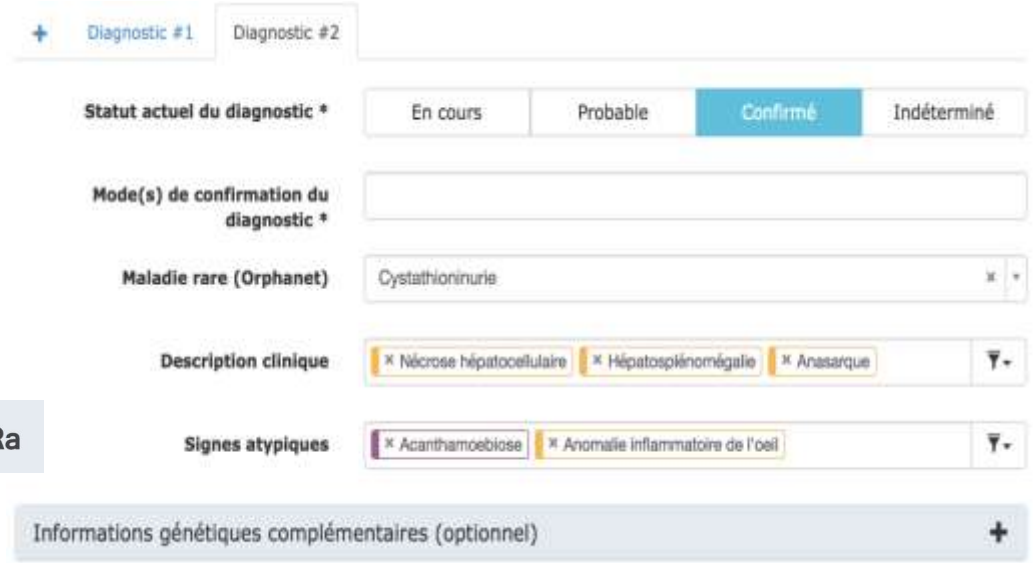
ORPHAcode	PreferredTerm	Synonyms	ICDcodes
166024	Multiple epiphyseal dysplasia-macrocephaly-facial dysmorphism syndrome		Q77.3
166024	Multiple epiphyseal dysplasia-macrocephaly-facial dysmorphism syndrome	Dysplasie épiphysaire multiple type Al-Gazali	
166024	Multiple epiphyseal dysplasia-macrocephaly-facial dysmorphism syndrome	Multiple epiphyseal dysplasia, Al-Gazali type	
58	Alexander disease		G93.8
58	Alexander disease	AxD	
166032	Multiple epiphyseal dysplasia-miniepiphyses syndrome		Q77.3
61	Alpha-mannosidosis		E77.1
61	Alpha-mannosidosis	Lysosomal alpha-D-mannosidase deficiency	
166029	Multiple epiphyseal dysplasia-severe proximal femoral dysplasia syndrome		Q77.3
166038	Metaphyseal chondrodysplasia, Kaitila type		Q78.5
93	Aspartylglucosaminuria		E77.1
93	Aspartylglucosaminuria	Aspartylglucosaminidase deficiency	
166035	Brachydactyly-short stature-retinitis pigmentosa syndrome		Q87.8
585	Multiple sulfatase deficiency		E75.2

Orphanet releases standardized ICD-10, ICD-11 and SNOMED-CT mappings to ensure quality and univocity of reporting

Guideline V

Track the patient's diagnostic pathway

Guideline 5 - Keep track, for each patient file, of the different ORPHAcodes and associated versions that were used to describe the patient's diagnostic pathway.



BaMaRa

It is important to keep track of the different codes that were used to describe the patient's disorder, to understand his/her diagnostic pathway through diagnostic codes refinements

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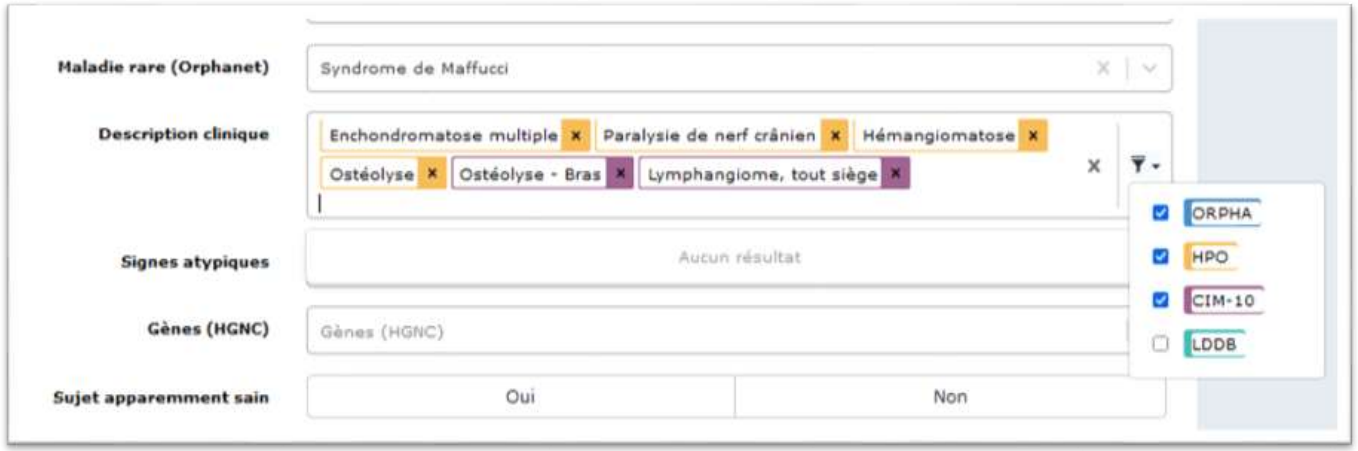


BaMaRa

ORPHAcodes can be combined with other descriptors (such as other codes), phenotypic traits (such as HPO clinical signs) and genotype information

Guideline IV

Resources updates

Guideline 4 - Update your coding resource according to the internationally agreed cycle annually in order to have the most recent coding file and to ensure comparability.

HIS (EHRs):

ANNUAL versioning Nomenclature Pack July

REGISTRIES

- **ANNUAL versioning (Nomenclature Pack)**
- **TWICE A YEAR (ORDO)**
- **TWICE A YEAR (Orphadata nomenclature and classifications files)**

http://www.rd-code.eu/wp-content/uploads/2022/01/826607_D5-4_Standard-procedure-and-guide-for-coding-with-Orphacodes_final.pdf

Coding support

REFER TO RD CODE > GUIDELINES & OD4RD(2) > HELPDESK

orphadata

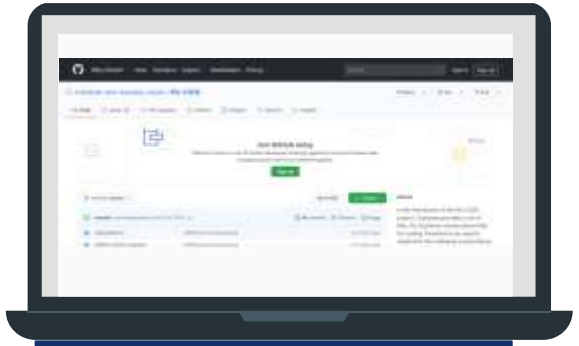
Orphanet nomenclature files for coding

>RD-CODE

OD4RD
Orphanet Data For Rare Diseases

www.orphadata.com

www.rd-code.eu



www.od4rd.eu
https://github.com/OD4RD

Some more useful resources

OD4RD GitHub
Wiki section
https://github.com/OD4RD/Main-Help-Desk/wiki

Online e-leanring course
developped by the Norway
Orphanet team
https://sjelden.no/nettkurs/orphacodes-for-rare-diseases/

Orphanet Reports
(Activity report)
https://www.orpha.net/en/other-information/reports

Orphanet standardized
procedures
https://www.orpha.net/en/other-information/procedures

ERDERA

European Rare Diseases Research Alliance

Thank you!

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