IMPLEMENTATION STORYLINE

OD4RD2 KoM

OD4RD
Orphanet Data For Rare Diseases

IMPLEMENTATION PROCESS

« DIY »
FULL
PROCESS

USING
SERVICES
(API)

- There is no « one for all » solutions
- Analyse your needs AND constrains
- Technical assessments and legal consideration are important

« DIY » : get the files !

« DIY » : get the files ! https://www.orphadata.com

HOME ORPHADATA SCIENCE SCIENTIFIC KNOWLEDGE BASE NOMENCLATURE FOR CODING EXPERT RESOURCES ONTOLOGIES TOOLS

Nomenclature pack 2022

Show 15 entries Search:

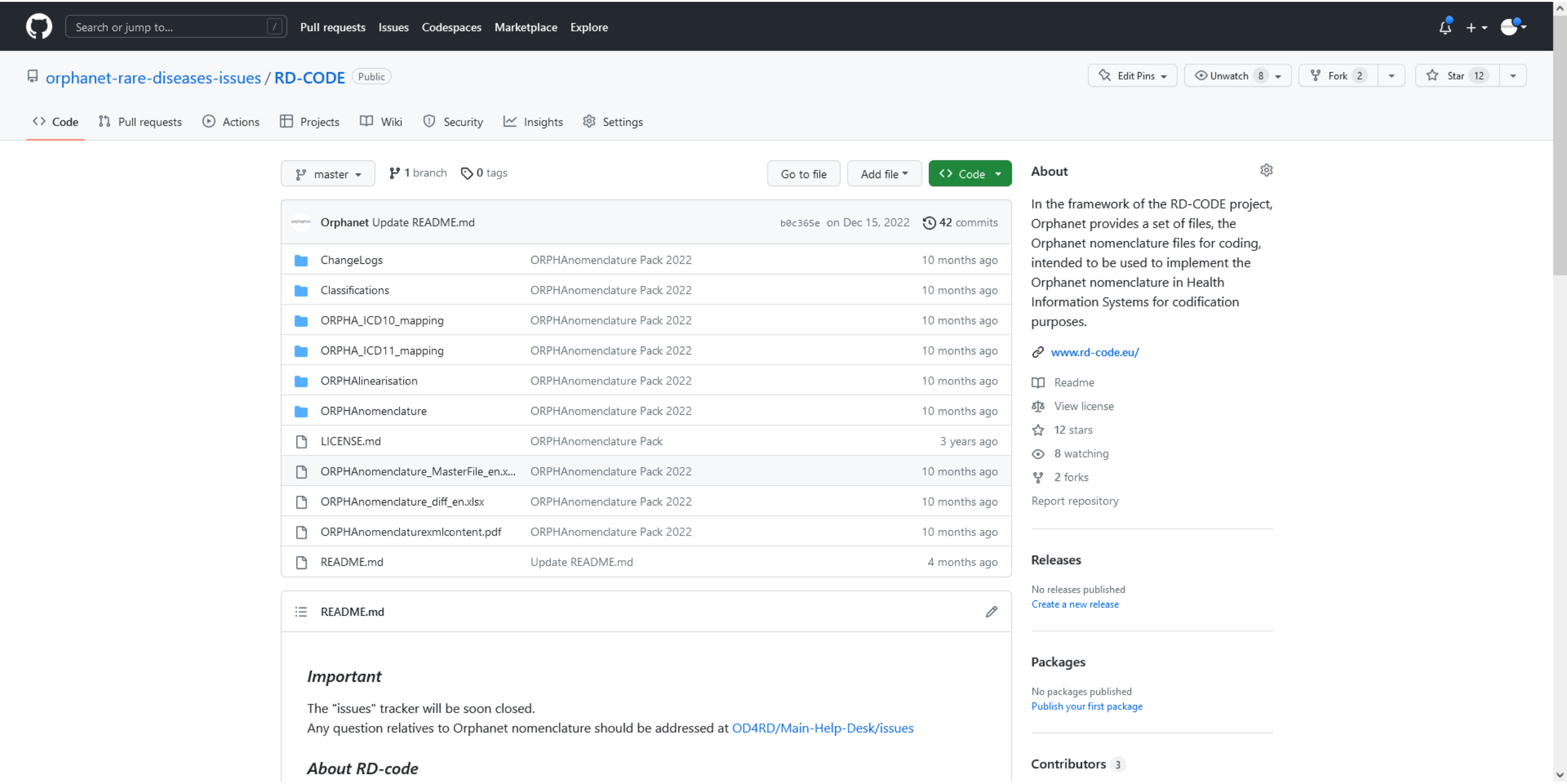
Language	File	Size
English	Orphanet_Nomenclature_Pack_EN.zip	5.61 MB
Czech	Orphanet_Nomenclature_Pack_CS.zip	4.64 MB
Dutch	Orphanet_Nomenclature_Pack_NL.zip	5.8 MB
French	Orphanet_Nomenclature_Pack_FR.zip	5.5 MB
German	Orphanet_Nomenclature_Pack_DE.zip	5.21 MB
Italian	Orphanet_Nomenclature_Pack_IT.zip	5.33 MB
Spanish	Orphanet_Nomenclature_Pack_ES.zip	5.71 MB
Polish	Orphanet_Nomenclature_Pack_PL.zip	5.07 MB
Portuguese	Orphanet_Nomenclature_Pack_PT.zip	4.64 MB

Showing 1 to 9 of 9 entries Previous Next

Previous nomenclature packs

Guidance for coding

« DIY » : get the files ! https://github.com/orphanet-rare-diseases-issues/RD-CODE

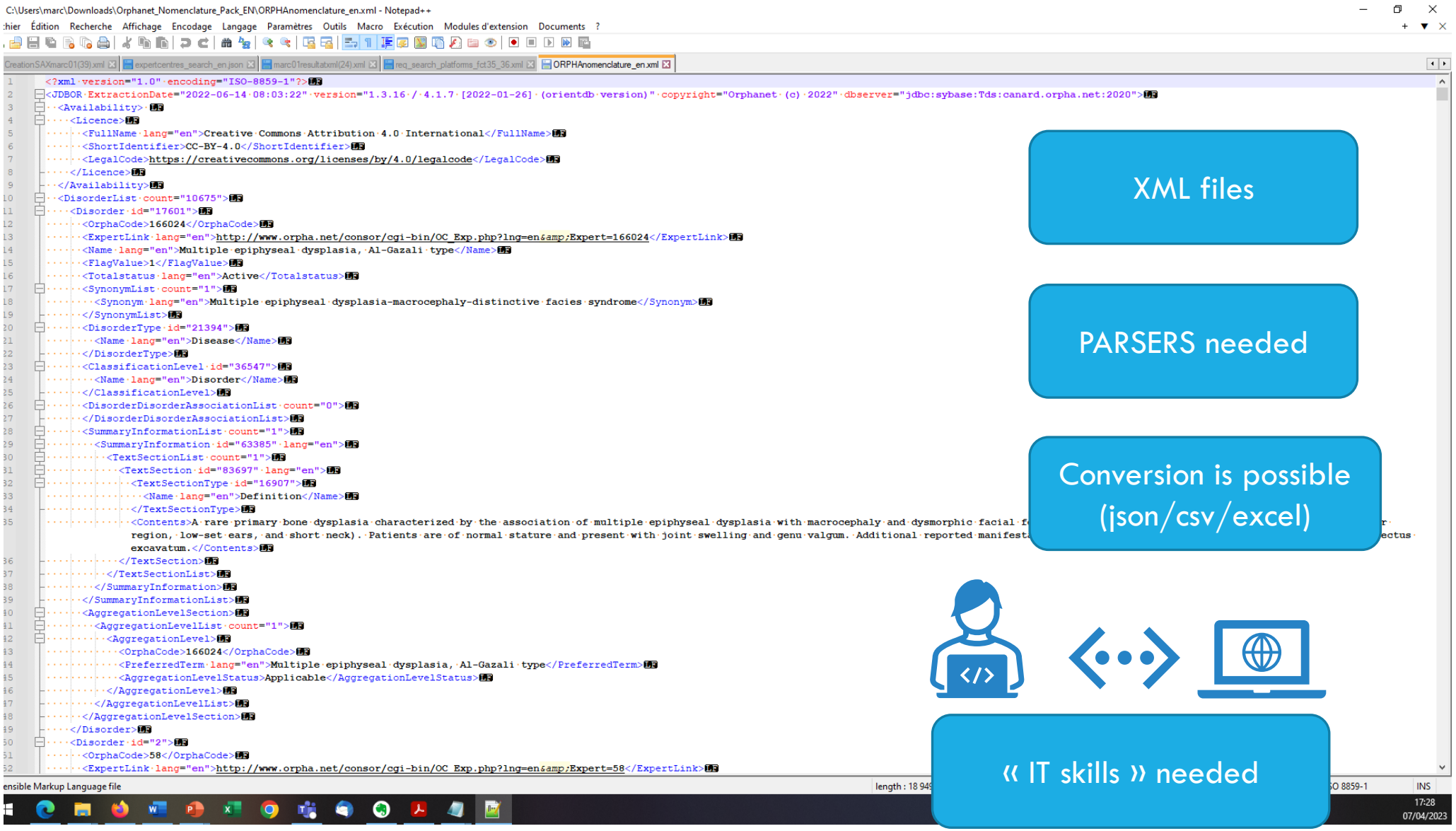


« DIY » : get the files ! https://www.orphadata.com

arc › Téléchargements › Orphanet_Nomenclature_Pack_EN

Nom	Modifié le	Type	Taille
Classifications	14/06/2022 08:18	Dossier de fichiers	
Orphanet_Nomenclature_Pack_en	04/10/2022 17:11	Dossier de fichiers	
ORPHA_ICD10_mapping_en.xml	14/06/2022 08:18	Document XML	8 994 Ko
ORPHA_ICD11_mapping_en.xml	14/06/2022 08:18	Document XML	1 223 Ko
ORPHAlinearisation_en.xml	14/06/2022 08:18	Document XML	5 681 Ko
ORPHAnomenclature_diff_en.xlsx	14/06/2022 08:50	Feuille de calcul ...	31 Ko
ORPHAnomenclature_en.xml	14/06/2022 08:18	Document XML	18 505 Ko
ORPHAnomenclature_MasterFile_2022.xlsx	14/06/2022 08:51	Feuille de calcul ...	529 Ko
ORPHAnomenclaturefilescontent_2022.pdf	07/06/2022 11:34	Document Adobe ...	596 Ko

« DIY » : get the files ! https://www.orphadata.com



XML files

PARSERS needed

Conversion is possible (json/csv/excel)

« IT skills » needed

Using API services ! (Application Programming Interfaces ARE made for this)

API FOR CODING RARE DISEASES

orphanet

>RD-CODE

ORPHA Nomenclature - RD-CODE project 2.2 OAS3

./openapi.json

The Orphanet nomenclature is a multilingual, standardised, controlled medical terminology specific to rare diseases, that includes all clinical entities registered in the Orphanet database (www.orpha.net). Each clinical entity (disorder, group of disorders, or subtype of a disorder) is associated with a unique numerical identifier named ORPHAcode, as well as a preferred term, synonyms, and a definition.

In the frame of the RD-CODE project, co-funded by the European Union's Third Health Program, Orphanet developped this API. It intends to support Member States in the implementation of the ORPHA nomenclature to rare diseases-specific codification systems. Starting with countries that have no systematic implementation of the Orpha codification yet, but that are actively committed already in doing so, this project will provide a sufficient real-world implementation experience to be captured by other countries in the future.

Disclaimers:

The API arise from the RD-CODE project which has received funding from the European Union in the framework of the Health Program.

The content of this API represents the views of the author only and is his/her sole responsibility; it cannot be considered to reflect the views of the European Commission and/or the Consumers, Health, Agriculture and Food Executive Agency or any other body of the European Union. The European Commission and the Agency do not accept any responsibility for use that may be made of the information it contains.

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Users:

In order to connect to this API, please click 'authorise' and enter a user name of your choosing

No SLA… but

Update: Once a year (july)

https://api.orphacode.org/

Using API services ! (Application Programming Interfaces ARE made for this)

API FOR CODING RARE DISEASES

All Orphanet clinical entities

GET `/{lang}/ClinicalEntity` Search for all Orphanet clinical entities

Preferred term

GET `/{lang}/ClinicalEntity/orphacode/{orphacode}/Name` Search for a clinical entity's preferred term by ORPHAcode

GET `/{lang}/ClinicalEntity/FindbyName/{label}` Search for a clinical entity by preferred term

GET `/{lang}/ClinicalEntity/ApproximateName/{label}` Search for a clinical entity by approximate preferred term

Synonym(s)

GET `/{lang}/ClinicalEntity/orphacode/{orphacode}/Synonym` Search for a clinical entity's synonym(s) by ORPHAcode

GET `/{lang}/ClinicalEntity/FindbyName/{label}/Synonym` Search for a clinical entity by synonym

GET `/{lang}/ClinicalEntity/ApproximateName/{label}/Synonym` Search for a clinical entity by approximate synonym

Definition

GET `/{lang}/ClinicalEntity/orphacode/{orphacode}/Definition` Search for a clinical entity's definition by ORPHAcode

Typology

GET `/{lang}/ClinicalEntity/orphacode/{orphacode}/Typology` Search for a clinical entity's typology by ORPHAcode.

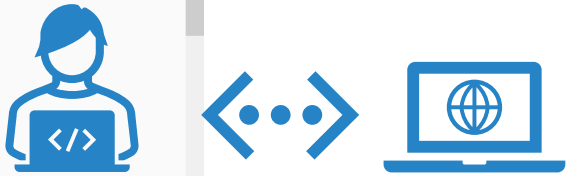
Orphanet URL

GET `/{lang}/ClinicalEntity/orphacode/{orphacode}/OrphanetURL` Search for a clinical entity's URL by ORPHAcode

Access to a full list of « queries »

Get what you need !

Build **your own backend**

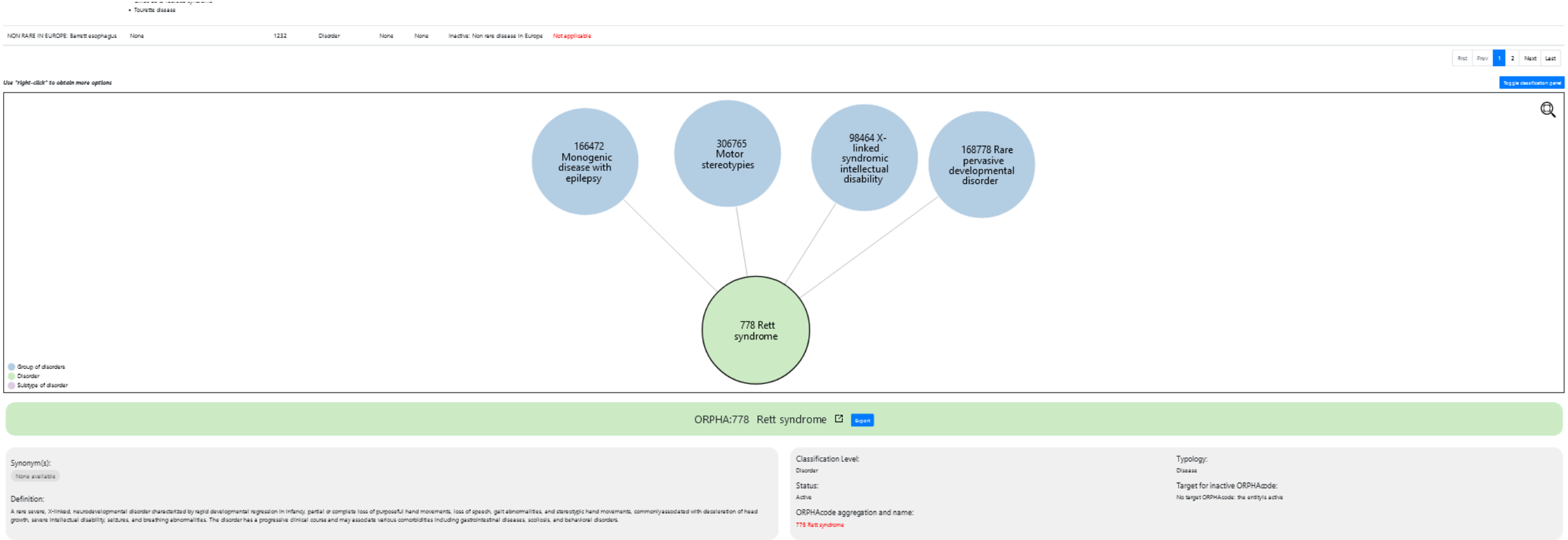


« IT skills » needed (but it's full standard JSON)

Don't use it for front-end directly

DATAVISUALISATION SERVICES

https://dataviz.orphacode.org/



MAPPING SERVICES

https://mappings.orphacode.org/

Mappor: Mapping orphacodes tool

Basic search | *Advanced search* | Search from input file

Disorder preferred term: Term(s) in disorder's preferred term...

Disorder synonym: Term(s) in disorder'synonym...

Disorder definition: microcephaly

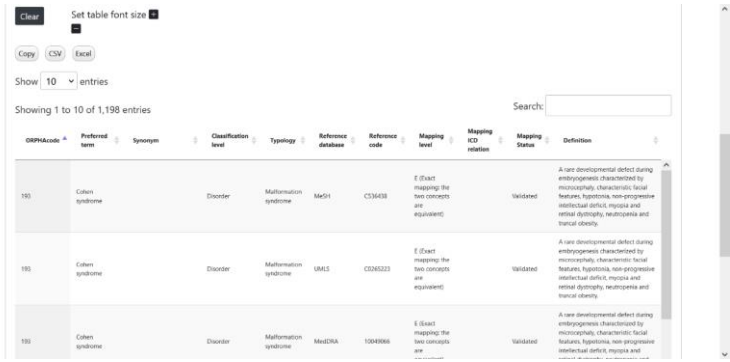
Reference database: Any

Reference code: You must select a reference database to enter a code...

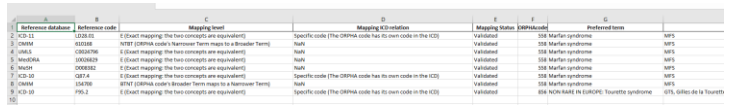
Mapping-level type: Any

Search | Clear all

Clear | Set table font size + −



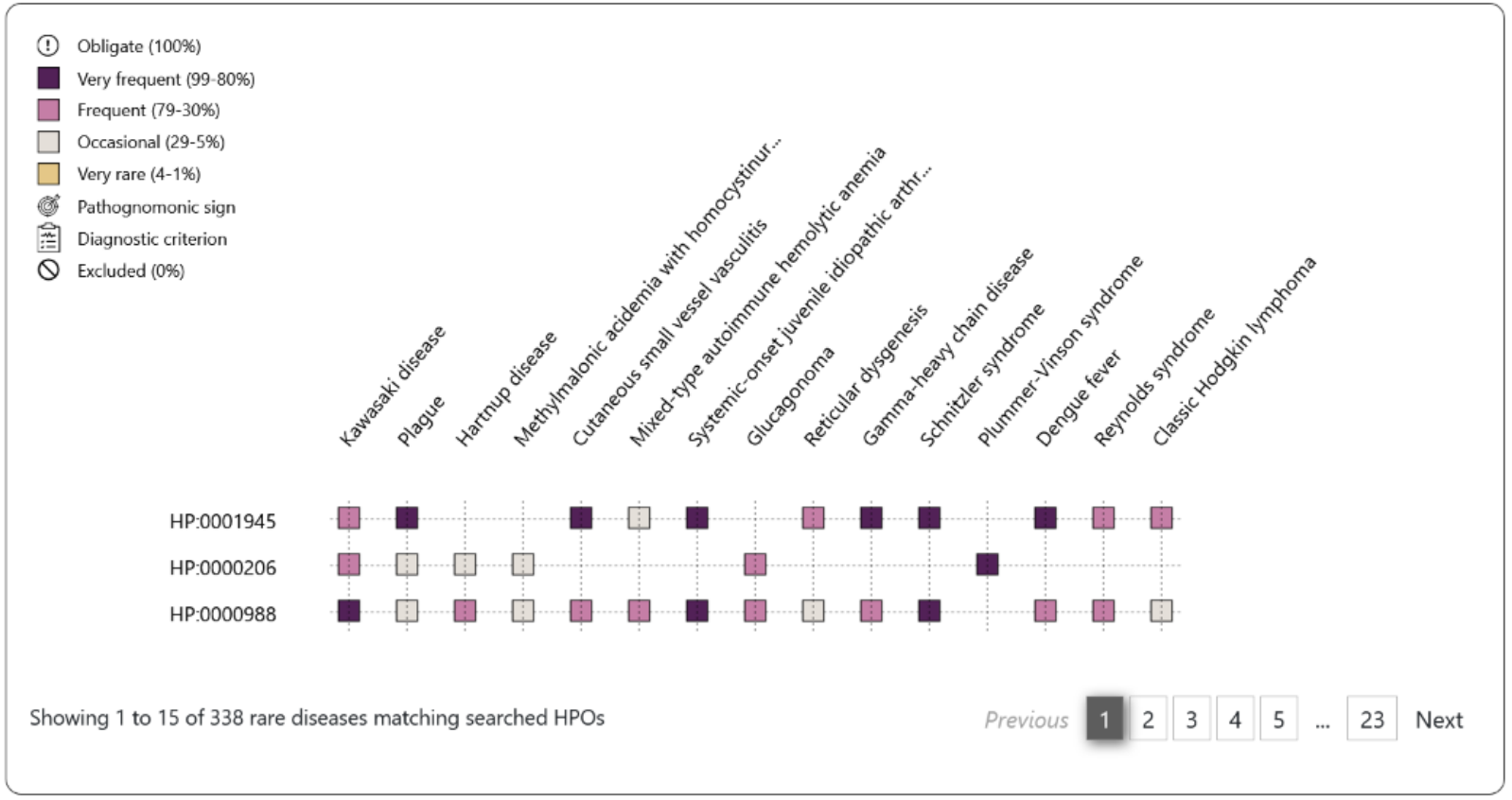
DOWNLOADABLE RESULTS



Csv/excel Files based mappings (input/output)

SEARCH BY CLINICAL SIGNS TOOLS (HPO)

https://clinicalsigns.orphanet.app/



API MANAGEMENT (GATEWAY).

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Next steps !

« legal » process support

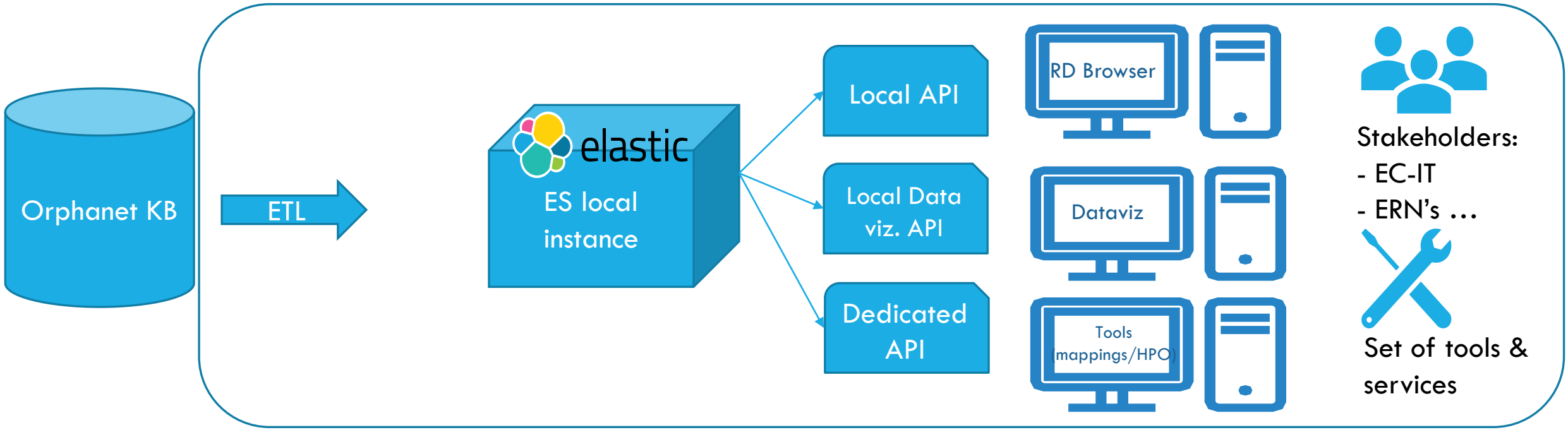
SLA !

SERVICES « AT SOURCE »

« legal » process support

Technical support

Local security assessment



DONT FORGET THE HELPDESK !

OD4RD2