

A complex network diagram with numerous nodes of varying sizes (dark blue, light blue, and grey) connected by thin grey lines. Some nodes are highlighted with larger concentric circles. The background is white with faint, larger-scale network patterns.

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 constraints
- ❁ Technical assessments and legal consideration are important

Orphanet Nomenclature Files for Coding



The Orphanet nomenclature pack compiles various files (listed below) 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 once a year, in 9 different languages: Czech, Dutch, English, French, German, Italian, Polish, Portuguese and Spanish. Excel and PDF files are available in English and are common to all Orphanet nomenclature pack.

Each year the Orphanet nomenclature pack includes :

- **Orphanet nomenclature file** ([XML Schema Definitions](#) and [JPEG](#) representations for this file).
- **Orphanet to ICD-10/ICD-11 mapping files** ([XML Schema Definitions](#) and [JPEG](#) representations for this files).
- **Orphanet classifications files by medical specialities** ([XML Schema Definitions](#) and [JPEG](#) representations for these files).

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

0 tags

Go to file

Add file ▾

<> Code ▾

About

In the framework of the RD-CODE project, Orphanet provides a set of files, the Orphanet nomenclature files for coding, intended to be used to implement the Orphanet nomenclature in Health Information Systems for codification purposes.

www.rd-code.eu/

 [Readme](#)

 [View license](#)

☆ 12 stars

8 watching

2 forks

Report repository

Releases

No releases published














[Create a new release](#)

Packages

No packages published

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Contributors 3

 Orphanet Update README.md		b0c365e on Dec 15, 2022	 42 commits
	ChangeLogs	ORPHAnomenclature Pack 2022	10 months ago
	Classifications	ORPHAnomenclature Pack 2022	10 months ago
	ORPHA_ICD10_mapping	ORPHAnomenclature Pack 2022	10 months ago
	ORPHA_ICD11_mapping	ORPHAnomenclature Pack 2022	10 months ago
	ORPHAlinearisation	ORPHAnomenclature Pack 2022	10 months ago
	ORPHAnomenclature	ORPHAnomenclature Pack 2022	10 months ago
	LICENSE.md	ORPHAnomenclature Pack	3 years ago
	ORPHAnomenclature_MasterFile_en.x...	ORPHAnomenclature Pack 2022	10 months ago
	ORPHAnomenclature_diff_en.xlsx	ORPHAnomenclature Pack 2022	10 months ago
	ORPHAnomenclaturexmlcontent.pdf	ORPHAnomenclature Pack 2022	10 months ago
	README.md	Update README.md	4 months ago

☰ README.md

Important

The "issues" tracker will be soon closed.

Any question relatives to Orphanet nomenclature should be addressed at [OD4RD/Main-Help-Desk/issues](#)

About RD-code

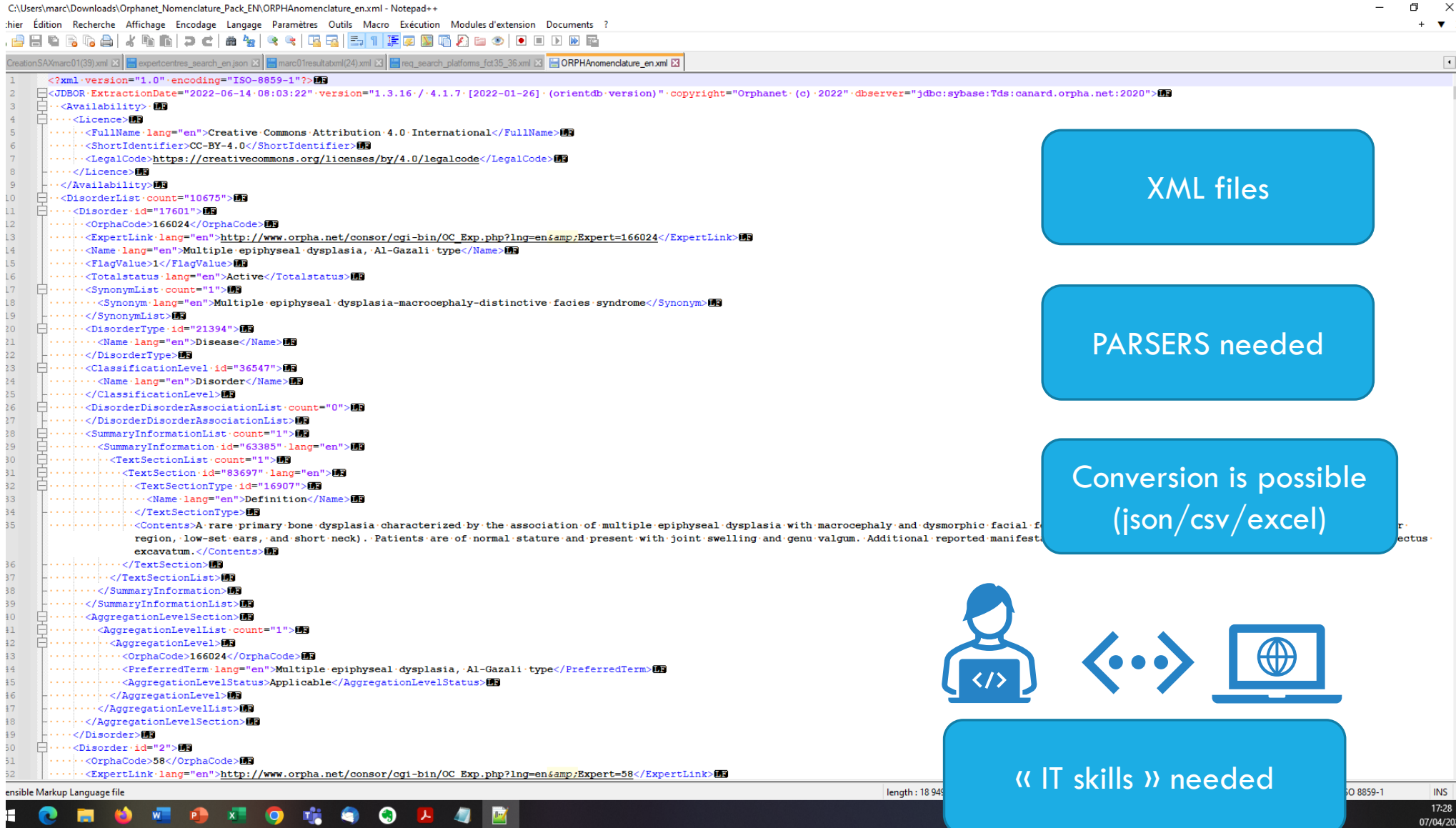


arc » Téléchargements » Orphanet_Nomenclature_Pack_EN

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

8859-1
INS
17:28
07/04/2023

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 ORPHAcodes, 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 developed 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

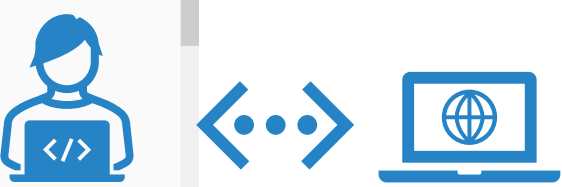


All Orphanet clinical entities			▼
GET	<code>/lang/ClinicalEntity</code>	Search for all Orphanet clinical entities	🔒
Preferred term			▼
GET	<code>/lang/ClinicalEntity/orphacode/{orphacode}/Name</code>	Search for a clinical entity's preferred term by ORPHAcode	🔒
GET	<code>/lang/ClinicalEntity/FindbyName/{label}</code>	Search for a clinical entity by preferred term	🔒
GET	<code>/lang/ClinicalEntity/ApproximateName/{label}</code>	Search for a clinical entity by approximate preferred term	🔒
Synonym(s)			▼
GET	<code>/lang/ClinicalEntity/orphacode/{orphacode}/Synonym</code>	Search for a clinical entity's synonym(s) by ORPHAcode	🔒
GET	<code>/lang/ClinicalEntity/FindbyName/{label}/Synonym</code>	Search for a clinical entity by synonym	🔒
GET	<code>/lang/ClinicalEntity/ApproximateName/{label}/Synonym</code>	Search for a clinical entity by approximate synonym	🔒
Definition			▼
GET	<code>/lang/ClinicalEntity/orphacode/{orphacode}/Definition</code>	Search for a clinical entity's definition by ORPHAcode	🔒
Typology			▼
GET	<code>/lang/ClinicalEntity/orphacode/{orphacode}/Typology</code>	Search for a clinical entity's typology by ORPHAcode.	🔒
Orphanet URL			▼
GET	<code>/lang/ClinicalEntity/orphacode/{orphacode}/OrphanetURL</code>	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/>

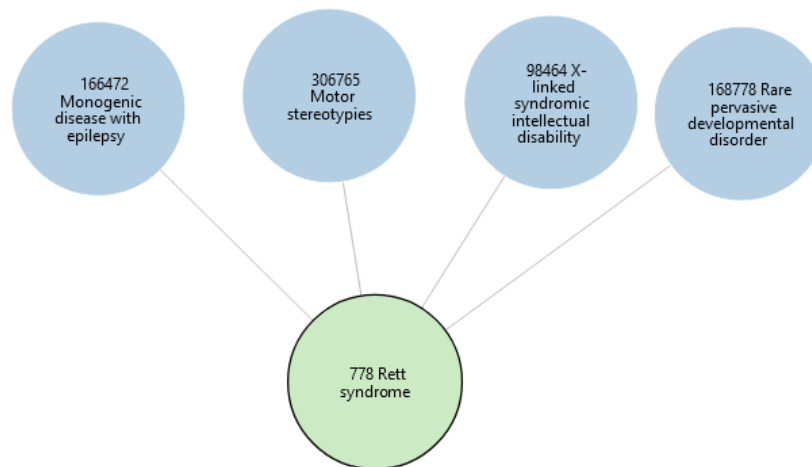
• Tourette disease

NON RARE IN EUROPE: Barrett esophagus None 1232 Disorder None None Inactive: Non rare disease in Europe Not applicable

First Prev 1 2 Next Last

Use "right-click" to obtain more options

Toggle classification pane



Group of disorders
Disorder
Subtype of disorder

ORPHA:778 Rett syndrome [Open](#)

Synonym(s):

None available

Definition:

A rare severe, X-linked, neurodevelopmental disorder characterized by rapid developmental regression in infancy, partial or complete loss of purposeful hand movements, loss of speech, gait abnormalities, and stereotypic hand movements, commonly associated with deceleration of head growth, severe intellectual disability, seizures, and breathing abnormalities. The disorder has a progressive clinical course and may associate various comorbidities including gastrointestinal diseases, scoliosis, and behavioral disorders.

Classification Level:

Disorder

Status:

Active

ORPHAcode aggregation and name:

778 Rett syndrome

Typology:

Disease

Target for inactive ORPHAcode:

No target ORPHAcode: the entity is active



MAPPING SERVICES

<https://mappings.orphacode.org/>

Mappor: Mapping orphacodes tool

Basic search

Advanced search

Search from input file

Disorder preferred term

Disorder synonym

Disorder definition

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

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

microcephaly

Reference database

Reference code

Mapping-level type

Any

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

Any

Search

Clear all

ORPHAcode	Preferred term	Synonym	Classification level	Typology	Reference database	Reference code	Mapping level	Mapping ICD relation	Mapping Status	Definition
103	Cohen syndrome		Disorder	Malformation syndrome	MeSH	C134438	E (Exact mapping: the two concepts are equivalent)		Validated	A rare developmental defect during embryogenesis characterized by microcephaly, characteristic facial features, hypotonia, non-progressive intellectual deficit, epilepsy and retinal dystrophy, neurocopia and trisomy 10.
185	Cohen syndrome		Disorder	Malformation syndrome	UMLS	0306023	E (Exact mapping: the two concepts are equivalent)		Validated	A rare developmental defect during embryogenesis characterized by microcephaly, characteristic facial features, hypotonia, non-progressive intellectual deficit, epilepsy and retinal dystrophy, neurocopia and trisomy 10.
103	Cohen syndrome		Disorder	Malformation syndrome	MeSH	0306023	E (Exact mapping: the two concepts are equivalent)		Validated	A rare developmental defect during embryogenesis characterized by microcephaly, characteristic facial features, hypotonia, non-progressive intellectual deficit, epilepsy and retinal dystrophy, neurocopia and trisomy 10.

DOWNLOADABLE RESULTS

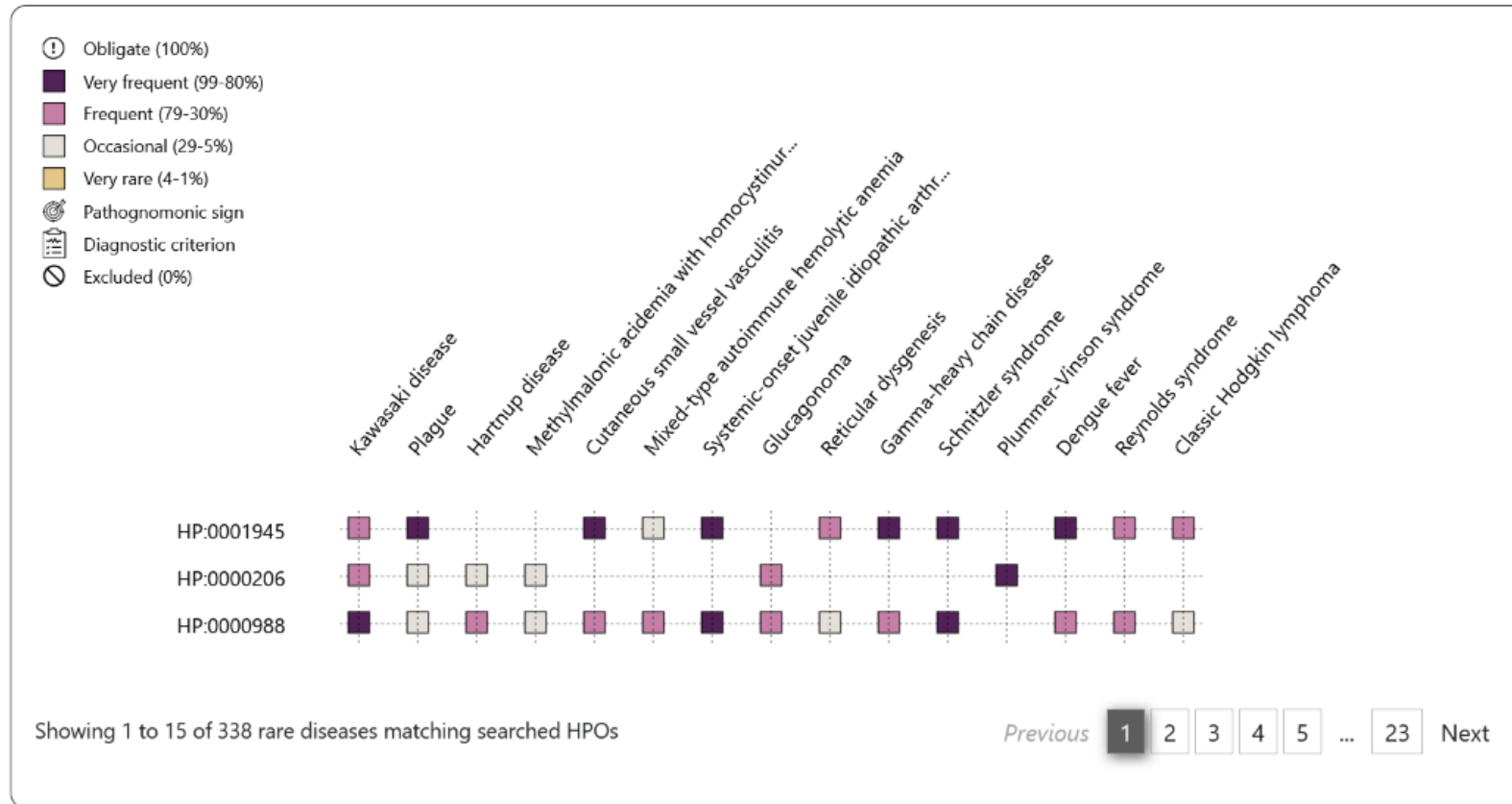
Reference database	Reference code	Mapping level	Mapping ICD relation	Mapping Status	Preferred term	Definition
ICD-11	1A2A.01	E (Exact mapping: the two concepts are equivalent)	Specific code (The ORPHA code has its own code in the ICD)	Validated	308 Marfan syndrome	MS
Orphanet	000000	WTF (ORPHA code's narrower term maps to a broader term)	None	Validated	308 Marfan syndrome	MS
UMLS	000000	E (Exact mapping: the two concepts are equivalent)	None	Validated	308 Marfan syndrome	MS
MeSH	000000	E (Exact mapping: the two concepts are equivalent)	None	Validated	308 Marfan syndrome	MS
ICD-10	Q87.4	E (Exact mapping: the two concepts are equivalent)	Specific code (The ORPHA code has its own code in the ICD)	Validated	308 Marfan syndrome	MS
ICD-11	1A2A.01	E (Exact mapping: the two concepts are equivalent)	Specific code (The ORPHA code has its own code in the ICD)	Validated	308 Marfan syndrome	MS
Orphanet	000000	WTF (ORPHA code's narrower term maps to a broader term)	None	Validated	308 Marfan syndrome	MS
ICD-10	P01.2	E (Exact mapping: the two concepts are equivalent)	Specific code (The ORPHA code has its own code in the ICD)	Validated	308 Marfan syndrome	MS

Csv/excel Files based mappings (input/output)



SEARCH BY CLINICAL SIGNS TOOLS (HPO)

<https://clinicalsigns.orphanet.app/>



API MANAGEMENT (GATEWAY).

orphanet

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Next
steps !

« legal » process
support

SLA !



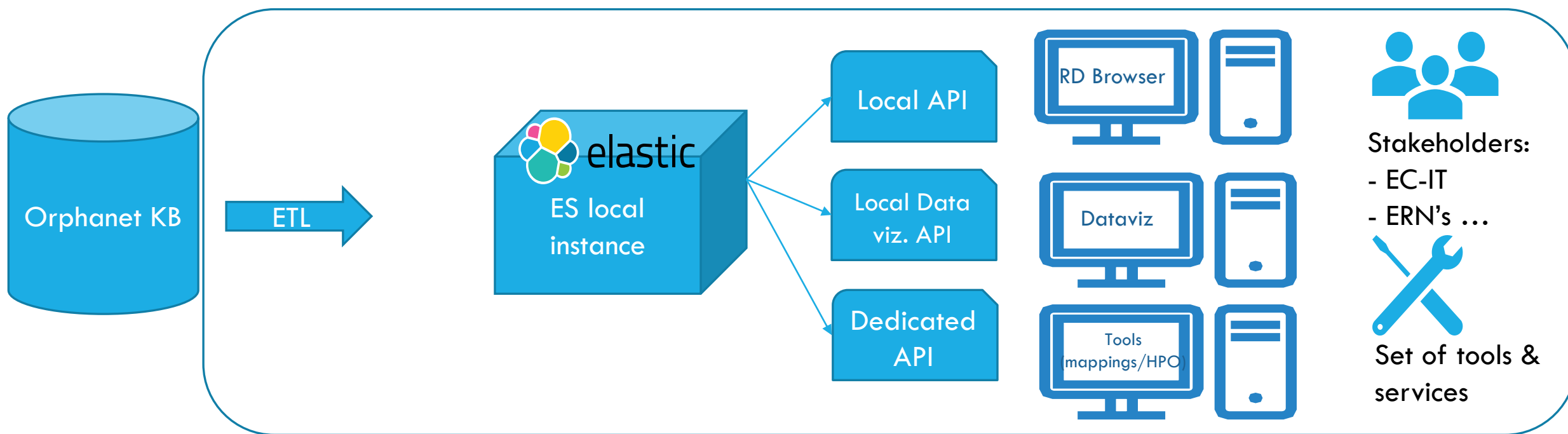
SERVICES « AT SOURCE »



« legal » process
support

Technical support

Local security
assessment



A complex network diagram with numerous nodes of varying sizes (dark blue, light blue, and grey) connected by thin grey lines. Some nodes are highlighted with larger concentric circles. The background is a light grey with faint, larger-scale network patterns.

DONT FORGET THE HELPDESK !

OD4RD2



OD4RD
Orphanet Data For Rare Diseases