



Orphacodes API« Application Programming Interface »

RD-CODE meeting

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Marc Hanauer,

acknowledgment: Daniel Murphy, Cyril Bigot, Valérie Lanneau, Annie Olry, Waed Abdel Khalek

★ <http://rd-code.eu>

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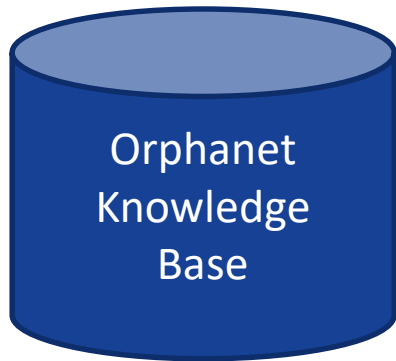
Glossary

- > API: Application Programming Interface
 - ★ Is a computing interface (not for « end users »)
 - ★ Defines interactions between « software » (I/O)
 - ★ It's a «grammar» explaining how to do « calls/queries » and expected results data format

- > JSON: JavaScript Object Notation
 - ★ It's a web standard
 - ★ language (programming) independent

- > XML: eXtensible Markup Language
 - ★ It's a standard for files and data exchanges
 - ★ Machine & Human readable

Data processing



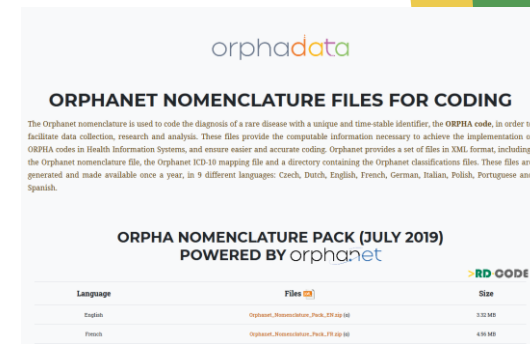
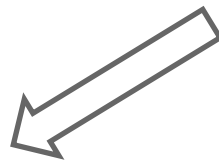
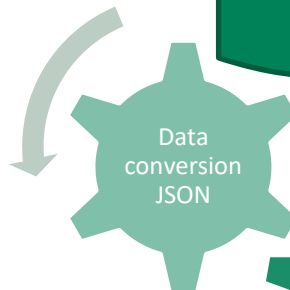
ElasticSearch
Cloud Instance



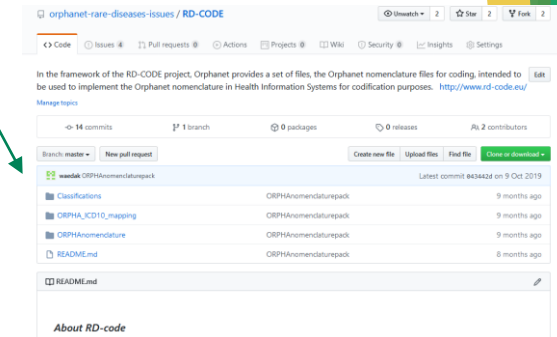
elastic



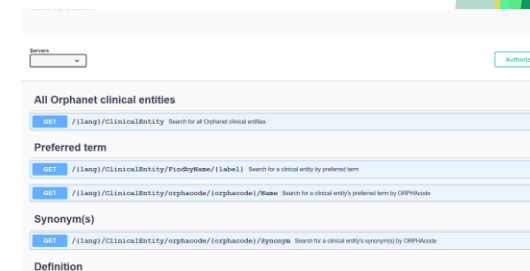
Nomenclature
Files
« Pack »
XML



Orphadata
Github



API





ElasticSearch Cloud Instance

- > ElasticSearch is a :
 - ★ Distributed [multitenant](#)-capable [full-text search](#) engine
 - ★ Schema Free, JSON based indexes
 - ★ Scalable solution, able to manage large amount of data
 - ★ Ready-to-use search API
 - ★ Back-end solution, not made for front or direct access
 - ★ Local instance or Cloud service (choice made)
 - ★ « Open core » business model (parts are open source, parts are proprietary such as rights/rules managements etc.)
- > RD-Code API
 - ★ As backend use the ElasticSearch API
 - ★ Reshape the JSON output from ElasticSearch to ease integration
 - ★ Insure « technical » independency (if we decide to use something else as backend) for the implementation side
 - ★ **Use a more standard way to describe itself**

RD-CODE API

- > OPEN API standard
 - Open API 3 swagger
 - <https://swagger.io/specification/>
 - <https://www.openapis.org/>
 - « State of the art » to design API
 - « Contract first » approach
 - Before coding the API the specifications are described in a standard way
 - Nice User interface ! Still human usable !
- > Only « GET » are allowed (you can't modify content...)



<https://api.orpha.net>

ORPHA Nomenclature - RD-CODE project

2.1-oas3

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 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.

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[Contact Orphanet team](#)

Servers



Authorize



<https://api.orphacode.org>

ORPHA Nomenclature - RD-CODE project 2.1-oas3 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, gene, drug, etc.) is associated with an ORPHAcode, as well as a preferred term, synonyms, and a definition.

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The content of this API represents the views of the European Union Member States in the implementation of the ORPHA nomenclature to rare diseases-specific information system (RD-CODE) already in doing so, this project will provide a standardised and structured information system.

We have chosen to apply the Commons Attribution-NonCommercial-ShareAlike license for the commercial use of these databases in all legislation.

Contact Orphanet team

Available authorizations

x

apiKey (apiKey)

Name: apiKey

In: header

Value:

test



Authorize

Close

Authorize





All Orphanet clinical entities	>
Preferred term	>
Synonym(s)	>
Definition	>
Typology	>
Orphanet URL	∨
<div><div>GET</div><div><code>/ {lang} / ClinicalEntity / orphancode / {orphancode} / OrphanetURL</code></div><div>Search for a clinical entity's URL by ORPHACode</div><div>🔒</div></div>	
Status	>
ORPHACode aggregation	>
Target for inactive ORPHACode	>
Classification level	>
Classification	>
Child	>
Parent	>
ICD10	>
OMIM	>

<https://api.orphacode.org>

All Orphanet clinical entities

{expected param}

GET [/{lang}/ClinicalEntity](#) Search for all Orphanet clinical entities

Preferred term

GET [/{lang}/ClinicalEntity/FindbyName/{label}](#) Search for a clinical entity by preferred term

GET [/{lang}/ClinicalEntity/orphacode/{orphacode}/Name](#) Search for a clinical entity's preferred term by ORPHAcode

Synonym(s)

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

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.

<https://api.orphacode.org>

All Orphanet clinical entities

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

The result is a collection of clinical entities (active/inactive) specifying the ORPHAcode, the preferred term, the status and the definition (when one exists) for each entity.

Parameters

Name	Description
------	-------------

lang * required

string

(path)

Language

Available values : CS, DE, EN, ES, FR, IT, NL, PL, PT

Default value : EN

EN

Responses

Code	Description
------	-------------

Links

<https://api.orphacode.org>

Responses

Code	Description	Links
200	<div>Dataset</div> <div>Media type</div> <div>application/json</div> <div>Controls Accept header.</div> <div>Example Value Schema</div> <pre>[{ "Date": "2019-07-10 11:47:17", "Definition": "A rare sex chromosome number anomaly disorder characterized, genetically, by the presence of an extra X and Y chromosome in males and, clinically, by tall stature, dysfunctional testes associated with infertility and insufficient testosterone production, cognitive, affective and social functioning impairments, global developmental delay, and an increased risk of congenital malformations.", "ORPHAcode": 10, "Preferred term": "48,XXYY syndrome", "Status": "Active" }]</pre>	No links
400	Bad request	No links
401	Authorization information is missing or invalid	No links
403		No links

<https://api.orphacode.org>

400	Bad request	No links
401	Authorization information is missing or invalid	No links
403	Bad server request	No links
404	Error	No links
	<div>Media type</div> <div>application/json ▾</div> <div>Example Value Schema</div> <div><pre>{ "code": 404, "message": "Error"}</pre></div>	
default	Unexpected error	No links
	<div>Media type</div> <div>application/json ▾</div> <div>Example Value Schema</div> <div><pre>{ "code": 404, "message": "Error"}</pre></div>	

<https://api.orphacode.org>

All Orphanet clinical entities

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

The result is a collection of clinical entities (active/inactive) specifying the ORPHACode, the preferred term, the status and the definition (when one exists) for each entity.

Parameters

Cancel

Name	Description
------	-------------

lang ★ required

string

(path)

Language

EN

Execute

Responses

<https://api.orphacode.org>

Curl

```
curl -X GET "https://api.orphacode.org/EN/ClinicalEntity" -H "accept: application/json" -H "apiKey: test"
```

Request URL

Request URL

<https://api.orphacode.org/EN/ClinicalEntity>

```
{
  "Preferred term": "Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency",
  "Status": "Active"
},
{
  "Date": "2019-07-24 16:05:36",
  "Definition": "3-methylcrotonyl-CoA carboxylase deficiency (3-MCCD) is an inherited disorder of leucine metabolism characterized by a highly variable clinical picture ranging from metabolic crisis in infancy to asymptomatic adults.",
  "ORPHAcode": "6",
  "Preferred term": "3-methylcrotonyl-CoA carboxylase deficiency",
  "Status": "Active"
},
{
  "Date": "2019-07-24 16:05:36",
  "Definition": "Cranio-cerebello-cardiac (3C) syndrome is a rare multiple congenital anomalies syndrome characterized by craniofacial (prominent occiput and forehead, hypertelorism, ocular coloboma, cleft palate), cerebellar (Dandy-Walker malformation, cerebellar vermis hypoplasia) and cardiac (tetralogy of Fallot,
```

<https://api.orphacode.org>

GET

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



The result retrieves the clinical entity's ORPHAcode and its preferred term.

Parameters

Cancel

Name	Description
------	-------------

lang * required

string
(path)

orphacode * required

integer
(path)

Request URL

```
https://api.orphacode.org/EN/ClinicalEntity/orphacode/558/Name
```

Server response

Code	Details
------	---------

200

Response body

```
{
  "Date": "2019-07-24 16:05:36",
  "ORPHAcode": "558",
  "Preferred term": "Marfan syndrome"
}
```

<https://api.orphacode.org>

<https://api.orphacode.org/EN/ClinicalEntity/orphacode/558/Name>

JSON	Données brutes	En-têtes
Enregistrer	Copier	Formater et indenter

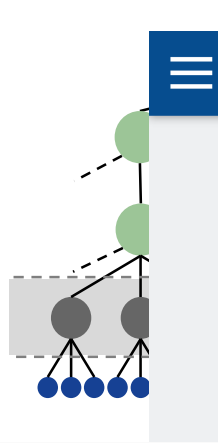
```
{  
  "Date": "2019-07-24 16:05:36",  
  "ORPHAcode": "558",  
  "Preferred term": "Marfan syndrome"  
}
```

(brower json interpreter)

JSON	Données brutes	En-têtes		
Enregistrer	Copier	Tout réduire	Tout développer	🔍 Filtrer le JSON

Date:	"2019-07-24 16:05:36"
ORPHAcode:	"558"
Preferred term:	"Marfan syndrome"

<https://api.orphacode.org>



orpha^{net}

Disease name

Marfan syndrome type 2

ORPHA:284973

[Classification level: Subtype of disorder](#)

Synonym(s): *Age of onset: All ages*

ORPHAcode aggregation

GET `/lang/ClinicalEntity/orphacode/{orphacode}/ORPHAcodeAggregation` Search for a clinical entity's aggregation code by ORPHAcode

The result retrieves the clinical entity's ORPHAcode, its preferred term as well as the ORPHAcode and the preferred term of the aggregation code.

Parameters

Name	Description
lang * required string (path)	Language
orphacode * required integer (path)	A unique and time-stable numerical identifier attributed randomly by the Orphanet database to each clinical entity upon its creation.

<https://api.orphacode.org>

ORPHACode aggregation

GET `/{{lang}}/ClinicalEntity/orphacode/{orphacode}/ORPHACodeAggregation` Search for a clinical entity's aggregation code by ORPHACode

The result retrieves the clinical entity's ORPHACode, its preferred term as well as the ORPHACode and the preferred term of the aggregation code.

Request URL

```
https://api.orphacode.org/EN/ClinicalEntity/orphacode/284973/ORPHACodeAggregation
```

Server response

Code	Details
------	---------

200	
-----	--

Response body

```
{
  "Date": "2019-07-24 16:05:36",
  "ORPHACodeAggregation": "558",
  "Preferred term": "Marfan syndrome"
}
```

<https://api.orphacode.org>

Child

GET

`/({lang})/Classification/{hchid}/orphacode/{orphacode}/Child` Search for a clinical entity's child(ren) by ORPHAcod and the unique identifier of a classification



Request URL

```
https://api.orphacode.org/EN/Classification/189/orphacode/558/Child
```

200

Response body

```
{
  "Child": [
    {
      "ORPHAcod": "284963",
      "Preferred term": "Marfan syndrome type 1"
    },
    {
      "ORPHAcod": "284973",
      "Preferred term": "Marfan syndrome type 2"
    }
  ],
  "Classification": {
    "ID of the classification": "189",
    "Name of the classification": "Rare ophthalmic disorder"
  },
  "Date": "2019-07-10 11:49:09",
  "ORPHAcod": "558",
  "Preferred term": "Marfan syndrome"
}
```

<https://api.orphacode.org>

Usage and SDK Samples

[Curl](#)[Java](#)[Android](#)[Obj-C](#)[JavaScript](#)[C#](#)[PHP](#)[Perl](#)[Python](#)

```
curl -X GET -H "api_key: [[apiKey]]" -H "Authorization: Basic [[basicHash]]" "https://virtserver.swaggerhub.com/orphanet/ClinicalEntity/2-oas3/{lang}/ClinicalEntity/orphannumber/{id}/Name"
```

```
from __future__ import print_statement
import time
import swagger_client
from swagger_client.rest import ApiException
from pprint import pprint

# Configure API key authorization: api_key
swagger_client.configuration.api_key['api_key'] = 'YOUR_API_KEY'
# Uncomment below to setup prefix (e.g. Bearer) for API key, if needed
# swagger_client.configuration.api_key_prefix['api_key'] = 'Bearer'
# Configure HTTP basic authorization: basic_auth
swagger_client.configuration.username = 'YOUR_USERNAME'
swagger_client.configuration.password = 'YOUR_PASSWORD'

# create an instance of the API class
api_instance = swagger_client.NameApi()
lang = lang_example # String | Desired language
id = 789 # Long | ORPHAnumber of entity

try:
    # Search for the preferred term of the clinical entity by its ORPHAnumber.
    api_response = api_instance.list_name(lang, id)
```

Questions and suggestions

Preguntas y sugerencias

Întrebări și sugestii

Mistoqsijiet u suggerimenti

Otázky a návrhy