

# DESCRIPTION OF THE ORPHANET NOMENCLATURE FILES FOR CODING

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The Orphanet nomenclature files for coding is a set of xml files that includes an Orphanet nomenclature file, an ICD-10 – Orphanet nomenclature mapping file and a directory containing all the Orphanet classifications files in every Orphanet language. This document describes in details the content of these files and the way to explore them.

## 1. Orphanet Nomenclature of Rare Diseases files

The Orphanet nomenclature is used to code the diagnosis of rare diseases with a specific identifier named the ORPHANumber or ORPHAcode in order to facilitate data collection, research and analysis. The Orphanet Nomenclature files contain all entities present in Orphanet:

- active entities for direct use;
- and inactive entities that no longer belong to the nomenclature and are no longer part of the Orphanet classifications. These entities are, at the date of the file's generation, considered as Obsolete, Deprecated or Non rare in Europe (See Appendix).

As the Orphanet nomenclature is organised in multi-hierarchical classifications, it might be difficult for unexperienced coders to identify the appropriate code. As recommended by the European Joint Action RD-Action, a reduced list of ORPHANumbers has been agreed upon in order to allow data sharing and statistical analysis at EU-level. This list, based on the disorder level, excluding groups and subtypes, establishes the “Aggregation level”.

This pack is available in different languages: English (EN), French (FR), Italian (IT), Dutch (NL), German (DE), Spanish (ES), Czech (CZ) and Polish (PL). This list may expand in the future.

For more definitions, please consult the [Appendix](#) of this document.

### 1.1 Description of the XML tags

- **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes of disorder) in the Xml file.
- **ORPHANumber:** Unique, time-stable and non-reusable numerical identifier. It is generated randomly by the database and named “ORPHAcode” when used for coding purposes. Currently, the ORPHANumber is made up of one to six digits. In the future, number of digits can expand.
- **ExpertLink:** stable URL pointing to the specific page of a given disease on the Orphanet website.
- **Name:** The most commonly accepted name in the medical community (preferred term), according to published consensus, opinion of experts of the relevant medical specialty and/or compelling predominance of the name in medical literature. Preferred terms are unique throughout the database, associated with one ORPHANumber only.
- **Totalstatus:** Status of the clinical entity. Can be either “Active”, “Inactive Deprecated”, “Inactive Obsolete”, “Inactive Non rare disease in Europe” or only one by clinical entity.
- **Flagvalue:** Flag value of the clinical entity.  
For “Active” entities: 1, 129 or 513  
For “Inactive deprecated” entities: 257  
For “Inactive obsolete” entities: 16 or 1024  
For “Inactive Non rare in Europe”: 48 or 1056
- **Synonym:** Terms that are perfectly equivalent to the preferred term. The number of synonyms is indefinite and may vary depending on the language of translation. Acronyms commonly used to describe the disease are included as synonyms.
- **GroupOfType:** High level type of the clinical entity. Can be either Group of disorders, disorder or subtype of disorder and only one by clinical entity.
- **DisorderType:** Type of the clinical entity. Can be either Category, Clinical group, Disease, Clinical syndrome, Malformation syndrome, Biological anomaly, Morphological anomaly, Particular clinical situation in a disease or syndrome, Etiological subtype, Clinical subtype or Histopathological subtype and only one by clinical entity.

- **DisorderDisorderAssociation:** Relationship between an inactive clinical entity and an active one advised to be used in replacement. Whenever possible, this association is provided for “Inactive Deprecated” and “Inactive Obsolete” clinical entities respectively with the AssociationType “Moved to” or “Referred to”. The Inactive entity is identified as “InDisorder” and the Active entity for replacement as “OutDisorder”.
- **TextualInformation:** Textual information available for the ORPHANumber. Only definitions are provided here.
- **AggregationLevel:** Recommended ORPHANumber in Europe for data sharing and statistical reporting. It is Applicable in the case of the Disorders (aggregation level are themselves), and the subtypes (aggregation level is the closest disorder in the classification). No aggregation level is provided for groups. Only one disorder is provided at AggregationLevel.

## 1.2 Examples

### 1.2.1 Active entity

#### Example 1: Disorder

```
<DisorderList count="10462">
```

10642 is the total number of clinical entities in this XML file

```
<Disorder id="3555">
  <OrphaNumber>5</OrphaNumber>
  <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=5</ExpertLink>
  <Name lang="en">Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency</Name>
```

The concerned clinical entity has 5 as its ORPHA number and Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency as its preferred term in English. Following [http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?lng=en&Expert=5](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=5) will open the page of the Orphanet website dedicated to the clinical entity.

```
<FlagValue>1</FlagValue>
<Totalstatus lang="en">Active</Totalstatus>
```

The entity is in use in this version of the Orphanet Nomenclature.

```
<SynonymList count="3">
  <Synonym lang="en">LCHAD deficiency</Synonym>
  <Synonym lang="en">LCHADD</Synonym>
  <Synonym lang="en">Long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency</Synonym>
```

The entity has three synonyms in English.

```
<GroupOfType id="36547">
  <Name lang="en">Disorder</Name>
</GroupOfType>
```

The entity is a disorder, not a group, not a subtype.

```
<DisorderType id="21394">
  <Name lang="en">Disease</Name>
</DisorderType>
```

The entity is a disease, not a Clinical syndrome, Malformation syndrome, Biological anomaly, Morphological anomaly or Particular clinical situation in a disease or syndrome.

```
<DisorderDisorderAssociationList count="0">
</DisorderDisorderAssociationList>
```

As this entity is active, no association is provided.

```
<TextualInformationList count="1">
  <TextualInformation id="12552" lang="en">
    <TextSectionList count="1">
      <TextSection id="81974" lang="en">
        <TextSectionType id="16907">
          <Name lang="en">Disease definition</Name>
        </TextSectionType>
        <Contents>A mitochondrial disorder of long chain fatty acid oxidation characterized in most patients by onset in
infancy/ early childhood of hypoketotic hypoglycemia, metabolic acidosis, liver disease, hypotonia and, frequently, cardiac
involvement with arrhythmias and/or cardiomyopathy.</Contents>
      </TextSection>
    </TextSectionList>
  </TextualInformation>
```

</TextualInformationList>

A definition in English is available.

```
<AggregationLevelSection>
  <AggregationLevelList count="1">
    <AggregationLevel>
      <OrphanNumberAggregation>5</OrphanNumberAggregation>
      <PreferredTerm lang="en">Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency</PreferredTerm>
      <AggregationLevelStatus>Applicable</AggregationLevelStatus>
    </AggregationLevel>
  </AggregationLevelList>
</AggregationLevelSection>
```

As this entity is a disorder, the ORPHANumber of the aggregation level is itself and the corresponding status is Applicable.

</Disorder>

## Example 2: Group of disorders

```
<Disorder id="12622">
  <OrphaNumber>95498</OrphaNumber>
  <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=95498</ExpertLink>
  <Name lang="en">Congenital anomaly of superior vena cava</Name>
```

The concerned clinical entity has 95498 as its ORPHA number and Congenital anomaly of superior vena cava as its preferred term in English. Following [http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Ing=en&Expert=95498](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=95498) will open the page of the Orphanet website dedicated to the clinical entity.

```
<FlagValue>1</FlagValue>
<Totalstatus lang="en">Active</Totalstatus>
```

The entity is in use in this version of the Orphanet Nomenclature.

```
<SynonymList count="2">
  <Synonym lang="en">Congenital anomaly of superior caval vein</Synonym>
  <Synonym lang="en">Congenital anomaly of the SVC</Synonym>
```

The entity has two synonyms in English.

```
<GroupOfType id="36540">
  <Name lang="en">Group of disorders</Name>
</GroupOfType>
```

The entity is a group, not a disorder, not a subtype.

```
<DisorderType id="36561">
  <Name lang="en">Category</Name>
</DisorderType>
```

The entity is a category, not a clinical group.

```
<DisorderDisorderAssociationList count="0">
</DisorderDisorderAssociationList>
```

As this entity is active, no association is provided.

```
<TextualInformationList count="0">
</TextualInformationList>
```

The definition is not available.

```
<AggregationLevelSection>
  <AggregationLevelList count="0"/>
  <OrphanNumberAggregation/>
  <PreferredTerm/>
  <AggregationLevelStatus>Not applicable</AggregationLevelStatus>
</AggregationLevelSection>
```

As this entity is a group, the ORPHANumber of the aggregation level does not exist.

</Disorder>

## Example 3: Subtype of disorders

```
<Disorder id="12650">
  <OrphaNumber>95626</OrphaNumber>
  <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=95626</ExpertLink>
  <Name lang="en">Acquired central diabetes insipidus</Name>
```

The concerned clinical entity has 95626 as its ORPHA number and Adured central diabetes insipidus as its preferred term in English. Following [http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?lng=en&Expert=95626](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=95626) will open the page of the Orphanet website dedicated to the clinical entity.

```
<FlagValue>1</FlagValue>
<Totalstatus lang="en">Active</Totalstatus>
```

The entity is in use in this version of the Orphanet Nomenclature.

```
<SynonymList count="2">
  <Synonym lang="en">Acquired CDI</Synonym>
  <Synonym lang="en">Acquired neurogenic diabetes insipidus</Synonym>
```

The entity has two synonyms in English.

```
<GroupOfType id="36554">
  <Name lang="en">Subtype of disorder</Name>
</GroupOfType>
```

The entity is a subtype, not a group, not a disorder.

```
<DisorderType id="21450">
  <Name lang="en">Clinical subtype</Name>
</DisorderType>
```

The entity is a clinical subtype, not an etiological subtype or a histopathological subtype.

```
<DisorderDisorderAssociationList count="1">
  <DisorderDisorderAssociation>
    <OutDisorder id="12650" cycle="true"/>
    <InDisorder id="12649">
      <OrphaNumber>95625</OrphaNumber>
      <Name lang="en">OBSOLETE: Posttraumatic diabetes insipidus</Name>
    </InDisorder>
    <DisorderDisorderAssociationType id="27341">
      <Name lang="en">Referred to</Name>
    </DisorderDisorderAssociationType>
  </DisorderDisorderAssociation>
</DisorderDisorderAssociationList>
```

The entity OBSOLETE: Posttraumatic diabetes insipidus has been obsoleted (InDisorder). Acquired neurogenic diabetes insipidus should be used instead (OutDisorder).

```
<TextualInformationList count="1">
  <TextualInformation id="39282" lang="en">
    <TextSectionList count="1">
      <TextSection id="54203" lang="en">
        <TextSectionType id="16907">
          <Name lang="en">Disease definition</Name>
        </TextSectionType>
      </TextSection>
    </TextSectionList>
  </TextualInformation>
</TextualInformationList>
```

<Contents>Acquired central diabetes insipidus (acquired CDI) is a subtype of central diabetes insipidus (CDI, see this term), characterized by polyuria and polydipsia, due to an idiopathic or secondary decrease in vasopressin (AVP) production.</Contents>

```
</TextSection>
</TextSectionList>
</TextualInformation>
</TextualInformationList>
```

A definition in English is available.

```
<AggregationLevelSection>
  <AggregationLevelList count="1">
    <AggregationLevel>
      <OrphanNumberAggregation>178029</OrphanNumberAggregation>
      <PreferredTerm lang="en">Central diabetes insipidus</PreferredTerm>
      <AggregationLevelStatus>Applicable</AggregationLevelStatus>
    </AggregationLevel>
  </AggregationLevelList>
</AggregationLevelSection>
```

As this entity is a subtype, the ORPHA number of the aggregation level is the closest disorder in the classification. Its ORPHA number is ORPHA:178029 and its preferred term in English is "Central diabetes insipidus".

```
</Disorder>
```

## 1.2.2 Inactive entity

### Example 1: Deprecated entity

```
<Disorder id="963">
  <OrphaNumber>670</OrphaNumber>
  <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=670</ExpertLink>
  <Name lang="en">PIBIDS syndrome</Name>
```

The concerned clinical entity has 670 as its ORPHA number and PIBIDS as its preferred term in English. Following [http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Ing=en&Expert=670](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=670) will open the page of the Orphanet website dedicated to the clinical entity.

```
  <FlagValue>257</FlagValue>
  <Totalstatus lang="en">Inactive Deprecated</Totalstatus>
```

The entity is inactive in this version of the Orphanet Nomenclature. It has been deprecated and became a part of another disorder present in the Orphanet nomenclature.

```
  <SynonymList count="2">
    <Synonym lang="en">Trichothiodystrophy type F</Synonym>
    <Synonym lang="en">Trichothiodystrophy-sun sensitivity syndrome</Synonym>
```

The entity has two synonyms in English.

```
  <GroupOfType id="36547">
    <Name lang="en">Disorder</Name>
  </GroupOfType>
```

The entity is a disorder, not a group, not a subtype.

```
  <DisorderType id="21394">
    <Name lang="en">Disease</Name>
  </DisorderType>
```

The entity is a disease, not a Clinical syndrome, Malformation syndrome, Biological anomaly, Morphological anomaly or Particular clinical situation in a disease or syndrome.

```
  <DisorderDisorderAssociationList count="1">
    <DisorderDisorderAssociation>
      <OutDisorder id="10319">
        <OrphaNumber>33364</OrphaNumber>
        <Name lang="en">Trichothiodystrophy</Name>
      </OutDisorder>
      <InDisorder id="963" cycle="true"/>
      <DisorderDisorderAssociationType id="21471">
        <Name lang="en">Moved to</Name>
      </DisorderDisorderAssociationType>
    </DisorderDisorderAssociation>
  </DisorderDisorderAssociationList>
```

There is an association between this inactive entity and an active one. The Inactive entity, identified as "InDisorder", is "moved to" the active entity ORPHA:33364 Trichothiodystrophy, identified as "OutDisorder".

```
  <TextualInformationList count="0">
  </TextualInformationList>
```

The definition is not available.

```
  <AggregationLevelSection>
    <AggregationLevelList count="1">
      <AggregationLevel>
        <OrphanNumberAggregation>33364</OrphanNumberAggregation>
        <PreferredTerm lang="en">Trichothiodystrophy</PreferredTerm>
        <AggregationLevelStatus>Applicable</AggregationLevelStatus>
      </AggregationLevel>
    </AggregationLevelList>
  </AggregationLevelSection>
```

As this entity is inactive, it cannot be used for data sharing and statistical reporting. As the ORPHA number of replacement ORPHA:33364 Trichothiodystrophy is of disorder level, it can be used instead.

```
</Disorder>
```

### Example 2: Obsolete entity

```
<Disorder id="8586">
```

```

<OrphaNumber>719</OrphaNumber>
<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC\_Exp.php?Ing=en&Expert=719</ExpertLink>
<Name lang="en">OBSOLETE: Pili canulati</Name>

```

The concerned clinical entity has 719 as its ORPHA number and Pili canulati as its preferred term in English. Following [http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Ing=en&Expert=719](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=719) will open the page of the Orphanet website dedicated to the clinical entity.

```

<FlagValue>16</FlagValue>
<Totalstatus lang="en">Inactive Obsolete</Totalstatus>

```

The entity is inactive in this version of the Orphanet Nomenclature. It is removed from the nomenclature for maintenance purposes.

```

<SynonymList count="0">

```

The entity has 0 synonyms.

```

<GroupOfType id="36547">
  <Name lang="en">Disorder</Name>
</GroupOfType>

```

The entity is a disorder, not a group, not a subtype.

```

<DisorderType id="21394">
  <Name lang="en">Disease</Name>
</DisorderType>

```

The entity is a disease, not a Clinical syndrome, Malformation syndrome, Biological anomaly, Morphological anomaly or Particular clinical situation in a disease or syndrome.

```

<DisorderDisorderAssociationList count="1">
  <DisorderDisorderAssociation>
    <OutDisorder id="1610">
      <OrphaNumber>1410</OrphaNumber>
      <Name lang="en">Uncombable hair syndrome</Name>
    </OutDisorder>
    <InDisorder id="8586" cycle="true"/>
    <DisorderDisorderAssociationType id="27341">
      <Name lang="en">Referred to</Name>
    </DisorderDisorderAssociationType>
  </DisorderDisorderAssociation>
</DisorderDisorderAssociationList>

```

There is an association between this inactive entity and an active one. The Inactive entity, identified as “InDisorder”, is “referred to” the active entity ORPHA:1410 Uncombable hair syndrome, identified as “OutDisorder”.

```

<TextualInformationList count="0">
</TextualInformationList>

```

The definition is not available.

```

<AggregationLevelSection>
  <AggregationLevelList count="1">
    <AggregationLevel>
      <OrphanumberAggregation>1410</OrphanumberAggregation>
      <PreferredTerm lang="en">Uncombable hair syndrome</PreferredTerm>
      <AggregationLevelStatus>Applicable</AggregationLevelStatus>
    </AggregationLevel>
  </AggregationLevelList>
</AggregationLevelSection>

```

As this entity is inactive, it cannot be used for data sharing and statistical reporting. As the ORPHA number of replacement ORPHA:1410 Uncombable hair syndrome is of disorder level, it can be used instead.

```

</Disorder>

```

### Example 3: Non rare in Europe

```

<Disorder id="3009">
  <OrphaNumber>1244</OrphaNumber>
  <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC\_Exp.php?Ing=en&Expert=1244</ExpertLink>
  <Name lang="en">NON RARE IN EUROPE: Bicuspid aortic valve</Name>

```

The concerned clinical entity has 1244 as its ORPHA number and Bicuspid aortic valve as its preferred term in English. Following [http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Ing=en&Expert=1244](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=1244) will open the page of the Orphanet website dedicated to the clinical entity.

```

<FlagValue>1056</FlagValue>

```



<Totalstatus lang="en">Inactive Non rare disease in Europe</Totalstatus>

The entity is inactive in this version of the Orphanet Nomenclature. It is regarded as a non rare disorder in this version of the Orphanet nomenclature with a point prevalence more than 1/2'000 in the general population.

<SynonymList count="0">

The entity has 0 synonyms.

<GroupOfType id="36547">

<Name lang="en">Disorder</Name>

</GroupOfType>

The entity is a disorder, not a group, not a subtype.

<DisorderType id="21415">

<Name lang="en">Morphological anomaly</Name>

</DisorderType>

The entity is a Morphological anomaly, not a disease, Clinical syndrome, Malformation syndrome, Biological anomaly, or Particular clinical situation in a disease or syndrome.

<DisorderDisorderAssociationList count="0">

</DisorderDisorderAssociationList>

As this entity is non rare in Europe, no association to a clinical entity in the Orphanet nomenclature can be provided.

<TextualInformationList count="0">

</TextualInformationList>

The definition is not available.

<AggregationLevelSection>

<AggregationLevelList count="0"/>

<OrphanumberAggregation/>

<PreferredTerm/>

<AggregationLevelStatus>Not applicable</AggregationLevelStatus>

</AggregationLevelSection>

This entity is inactive because it is non rare. Data sharing and statistical reporting in Europe should not be set out in association with an ORPHANumber. Accordingly, no aggregation level is available.

</Disorder>

#### Example 4: Inactive entity referred to a group of disorders

<Disorder id="8773">

<OrphaNumber>28455</OrphaNumber>

<ExpertLink lang="en">[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Ing=en&Expert=28455](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=28455)</ExpertLink>

<Name lang="en">OBSOLETE: Pancreatic beta cell agenesis with neonatal diabetes mellitus</Name>

The concerned clinical entity has 28455 as its ORPHA number and Pancreatic beta cell agenesis with neonatal diabetes mellitus as its preferred term in English. Following [http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Ing=en&Expert=28455](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=28455) will open the page of the Orphanet website dedicated to the clinical entity.

<FlagValue>16</FlagValue>

<Totalstatus lang="en">Inactive Obsolete</Totalstatus>

The entity is inactive in this version of the Orphanet Nomenclature. It is removed from the nomenclature for maintenance purposes.

<SynonymList count="0">

The entity has 0 synonyms.

<GroupOfType id="36547">

<Name lang="en">Disorder</Name>

</GroupOfType>

The entity is a disorder, not a group, not a subtype.

<DisorderType id="21394">

<Name lang="en">Disease</Name>

</DisorderType>

The entity is a disease, not a Clinical syndrome, Malformation syndrome, Biological anomaly, Morphological anomaly or Particular clinical situation in a disease or syndrome.

<DisorderDisorderAssociationList count="1">

<DisorderDisorderAssociation>

<OutDisorder id="18292">

<OrphaNumber>183625</OrphaNumber>

<Name lang="en">Rare genetic diabetes mellitus</Name>

```

</DisorderOut>
<InDisorder id="8773" cycle="true"/>
<DisorderDisorderAssociationType id="27341">
  <Name lang="en">Referred to</Name>
</DisorderDisorderAssociationType>
</DisorderDisorderAssociation>
</DisorderDisorderAssociationList>

```

There is an association between this inactive entity and an active one. The Inactive entity, identified as “InDisorder”, is “referred to” the active entity ORPHA:183625 Rare genetic diabetes mellitus, identified as “OutDisorder”.

```

<TextualInformationList count="0">
</TextualInformationList>

```

The definition is not available.

```

<AggregationLevelSection>
  <AggregationLevelList count="0"/>
  <OrphanNumberAggregation/>
  <PreferredTerm/>
  <AggregationLevelStatus>Not applicable</AggregationLevelStatus>
</AggregationLevelSection>

```

As this entity is inactive, it cannot be used for data sharing and statistical reporting. As the ORPHANumber of replacement ORPHA:183625 Rare genetic diabetes mellitus is of Group level, it cannot be used instead. The ORPHANumber of the aggregation level is not available.

```

</Disorder>

```

### Example 5: Inactive entity referred to a subtype

```

<Disorder id="12442">
  <OrphaNumber>93609</OrphaNumber>
  <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=93609</ExpertLink>
  <Name lang="en">Autosomal recessive distal renal tubular acidosis without deafness</Name>

```

The concerned clinical entity has 93609 as its ORPHA number and Autosomal recessive distal renal tubular acidosis without deafness as its preferred term in English. Following [http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Ing=en&Expert=93609](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=93609) will open the page of the Orphanet website dedicated to the clinical entity.

```

  <FlagValue>257</FlagValue>
  <Totalstatus lang="en">Inactive Deprecated</Totalstatus>

```

The entity is inactive in this version of the Orphanet Nomenclature. It becomes a part of another disorder present in the database.

```

  <SynonymList count="5">
    <Synonym lang="en">AR dRTA without deafness</Synonym>
    <Synonym lang="en">AR dRTA without hearing loss</Synonym>
    <Synonym lang="en">Autosomal recessive distal renal tubular acidosis without hearing loss</Synonym>
    <Synonym lang="en">Distal renal tubular acidosis type 1c</Synonym>
    <Synonym lang="en">dRTA type 1c</Synonym>
  </SynonymList>

```

The entity has 5 synonyms.

```

  <GroupOfType id="36554">
    <Name lang="en">Subtype of disorder</Name>
  </GroupOfType>

```

The entity is a subtype, not a group, not a disorder.

```

  <DisorderType id="21450">
    <Name lang="en">Clinical subtype</Name>
  </DisorderType>

```

The entity is a clinical subtype, not an etiological subtype or a histopathological subtype.

```

  <DisorderDisorderAssociationList count="1">
    <DisorderDisorderAssociation>
      <OutDisorder id="22876">
        <OrphaNumber>402041</OrphaNumber>
        <Name lang="en">Autosomal recessive distal renal tubular acidosis</Name>
      </OutDisorder>
      <InDisorder id="12442" cycle="true"/>
      <DisorderDisorderAssociationType id="21471">
        <Name lang="en">Moved to</Name>
      </DisorderDisorderAssociationType>
    </DisorderDisorderAssociation>
  </DisorderDisorderAssociationList>

```

```

</DisorderDisorderAssociationType>
</DisorderDisorderAssociation>
</DisorderDisorderAssociationList>

```

There is an association between this inactive entity and an active one. The inactive entity, identified as "InDisorder", is "moved to" the active entity ORPHA:402041 Autosomal recessive distal renal tubular acidosis, identified as "OutDisorder".

```

<TextualInformationList count="0">
</TextualInformationList>

```

The definition is not available.

```

<AggregationLevelSection>
<AggregationLevelList count="1">
<AggregationLevel>
<OrphanNumberAggregation>18</OrphanNumberAggregation>
<PreferredTerm lang="en">Distal renal tubular acidosis</PreferredTerm>
<AggregationLevelStatus>Applicable</AggregationLevelStatus>
</AggregationLevel>
</AggregationLevelList>
</AggregationLevelSection>

```

As this entity is inactive, it cannot be used for data sharing and statistical reporting. As the ORPHANumber of replacement ORPHA:402041 Autosomal recessive distal renal tubular acidosis is of subtype level, it cannot be used instead. The closest ORPHANumber of disorder level in the classification of ORPHA:402041 Autosomal recessive distal renal tubular acidosis is ORPHA:18 Distal renal tubular acidosis. It is used for aggregation level.

```

</Disorder>

```

## 2. Orphanet - ICD-10 cross referencing file

Orphanet maintains an alignment between ICD-10 (10th International Classification of Diseases established by the WorldHealth Organization - <https://icd.who.int/browse10/2016/en>) and its nomenclature of Rare Diseases.

The Orphanet - ICD-10 cross referencing file provides the ICD-10 code assigned to each ORPHANumber according to the [Orphanet ICD-10 coding rules](#). The mapping includes a semantic link that specifies the relationship between an ORPHANumber and an ICD-10 code.

For more definitions, please consult the [Appendix](#) of this document.

### 2.1 Description of the XML tags

- **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes of disorder) in the Xml file
- **ORPHANumber:** Unique, time-stable and non-reusable numerical identifier. It is generated randomly by the database and named "ORPHACode" when used for coding purposes
- **ExpertLink:** stable URL pointing to the specific page of a given disease on the Orphanet website.
- **Name:** The most commonly accepted name in the medical community (preferred term), according to published consensus, opinion of experts of the relevant medical specialty and/or compelling predominance of the name in medical literature. Preferred terms are unique throughout the database, associated to one ORPHANumber only.
- **Synonym:** Terms that are perfectly equivalent to the preferred term. The number of synonyms is indefinite and may vary depending on the language of translation. Acronyms commonly used to describe the disease are included as synonyms.
- **ExternalReferenceList:** list of cross-references for a given clinical entity in the ICD-10
- **DisorderMappingRelation:** Closeness relationship between an ORPHANumber and an ICD-10 code. Can be either E (Exact), NTBT (Narrow term to broad term), BTNT (Broad term to narrow term), ND (Not yet decided/unable to decide), W (Wrong) (See Appendix 2).

- **DisorderMappingICDRelation (ICD10 relationship):** Specificity relationship between an ORPHA number and ICD-10 code. Can be either Specific code, Inclusion term, Index term or Attributed (See Appendix 2).
- **DisorderMappingValidationStatus:** Validation status of the mapping between the ORPHA number and the ICD-10 code. Can be either Validated (mapping considered to be definite) or Not yet validated (provisional mapping needing further medical expertise).

## 2.2 Examples

### Example 1: ORPHA number has a specific code in ICD10

`<Disorder id="109">`

```
<OrphaNumber>558</OrphaNumber>
<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=558</ExpertLink>
<Name lang="en">Marfan syndrome</Name>
```

The concerned clinical entity has 558 as its ORPHA number and Marfan syndrome as its preferred term in English. Following [http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Ing=en&Expert=558](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=558) will open the page of the Orphanet website dedicated to the clinical entity.

```
<SynonymList count="1">
  <Synonym lang="en">MFS</Synonym>
</SynonymList>
```

The entity has one synonym.

```
<ExternalReferenceList count="1">
  <ExternalReference id="104868">
    <Source>ICD-10</Source>
    <Reference>Q87.4</Reference>
    <DisorderMappingRelation id="21527">
      <Name lang="en">E (exact mapping (the terms and the concepts are equivalent))</Name>
    </DisorderMappingRelation>
    <DisorderMappingICDRelation id="21583">
      <Name lang="en">Specific code (The term has its own code in the ICD10)</Name>
    </DisorderMappingICDRelation>
    <DisorderMappingValidationStatus id="21611">
      <Name lang="en">Validated</Name>
    </DisorderMappingValidationStatus>
  </ExternalReference>
</ExternalReferenceList>
```

This clinical entity is exactly mapped with ICD10 reference "Q87.4". This entity has its own code in ICD10. The relation between the reference and the clinical entity is "Validated".

`</Disorder>`

### Example 2: ORPHA number has an inclusion term on ICD10

`<Disorder id="106">`

```
<OrphaNumber>803</OrphaNumber>
<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=803</ExpertLink>
<Name lang="en">Amyotrophic lateral sclerosis</Name>
```

The concerned clinical entity has 803 as its ORPHA number and Amyotrophic lateral sclerosis as preferred term in English. Following [http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Ing=en&Expert=803](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=803) will open the page of the Orphanet website dedicated to the clinical entity.

```
<SynonymList count="3">
  <Synonym lang="en">ALS</Synonym>
  <Synonym lang="en">Charcot disease</Synonym>
  <Synonym lang="en">Lou Gehrig disease</Synonym>
</SynonymList>
```

The entity has three synonyms.

```
<ExternalReferenceList count="1">
  <ExternalReference id="104856">
    <Source>ICD-10</Source>
    <Reference>G12.2</Reference>
    <DisorderMappingRelation id="21534">
      <Name lang="en">NTBT (narrower term maps to a broader term)</Name>
    </DisorderMappingRelation>
```

```

<DisorderMappingICDRelation id="21590">
  <Name lang="en">Inclusion term (The term is included under a ICD10 category and has not its own code)</Name>
</DisorderMappingICDRelation>
<DisorderMappingValidationStatus id="21611">
  <Name lang="en">Validated</Name>
</DisorderMappingValidationStatus>
</ExternalReference>
</ExternalReferenceList>

```

This entity is mapped with ICD-10 reference "G12.2". It is a narrower term that maps to the broader term of "G12.2". The term is included in ICD-10 but under an ICD-10 category and does not have its own code. The relation between the reference and the clinical entity is "Validated".

```

</Disorder>

```

### Example 3: ORPHANumber has an index term on ICD10

```

<Disorder id="548">
  <OrphaNumber>635</OrphaNumber>
  <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=635</ExpertLink>
  <Name lang="en">Neuroblastoma</Name>

```

The concerned clinical entity has 635 as its ORPHA number and Neuroblastoma as its preferred term in English. Following [http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Ing=en&Expert=635](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=635) will open the page of the Orphanet website dedicated to the clinical entity.

```

  <SynonymList count="0">
  </SynonymList>

```

The entity has 0 synonyms.

```

  <ExternalReferenceList count="1">
    <ExternalReference id="106198">
      <Source>ICD-10</Source>
      <Reference>C74.9</Reference>
      <DisorderMappingRelation id="21534">
        <Name lang="en">NTBT (narrower term maps to a broader term)</Name>
      </DisorderMappingRelation>
      <DisorderMappingICDRelation id="21597">
        <Name lang="en">Index term</Name>
      </DisorderMappingICDRelation>
      <DisorderMappingValidationStatus id="21611">
        <Name lang="en">Validated</Name>
      </DisorderMappingValidationStatus>
    </ExternalReference>
  </ExternalReferenceList>

```

This entity is mapped with ICD-10 reference "C74.9". It is a narrower term that maps to the broader term of "C74.9". The term is matched at the ICD index term level and does not have its own code. The relation between the reference and the clinical entity is "Validated".

```

</Disorder>

```

### Example 4: ORPHANumber has an ICD10 term attributed by Orphanet

```

</Disorder>
<Disorder id="553">
  <OrphaNumber>2746</OrphaNumber>
  <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=2746</ExpertLink>
  <Name lang="en">Opsismodysplasia</Name>

```

The concerned clinical entity has 2746 as its ORPHA number and Opsismodysplasia as its preferred term in English. Following [http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Ing=en&Expert=2746](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=2746) will open the page of the Orphanet website dedicated to the clinical entity.

```

  <SynonymList count="0">
  </SynonymList>

```

The entity has 0 synonyms.

```

  <ExternalReferenceList count="1">
    <ExternalReference id="106212">
      <Source>ICD-10</Source>
      <Reference>Q78.8</Reference>
      <DisorderMappingRelation id="21534">
        <Name lang="en">NTBT (narrower term maps to a broader term)</Name>
      </DisorderMappingRelation>

```

```

<DisorderMappingICDRelation id="21604">
  <Name lang="en">Attributed</Name>
</DisorderMappingICDRelation>
<DisorderMappingValidationStatus id="21611">
  <Name lang="en">Validated</Name>
</DisorderMappingValidationStatus>
</ExternalReference>
</ExternalReferenceList>

```

This entity is mapped with ICD-10 reference "Q78.8". It is a narrower term that maps to the broader term of "Q78.8". The ORPHA clinical entity has no matching term at all in ICD-10. The ICD-10 code is attributed by Orphanet. The relation between the reference and the clinical entity is "Validated".

```
</Disorder>
```

### 3. Orphanet rare disease classification files

The Orphanet nomenclature is classified by medical specialties to reflect the multidimensional nature of rare diseases. Every entity can belong to multiple specialties according to their clinical presentation, and so be included in several classifications. The production and update of the classifications are based on scientific publications in peer-reviewed journals and in consultation with internationally identified experts. Only active clinical entities are part of the classifications.

The classification repository is available in the various Orphanet languages (English (EN), French (FR), Italian (IT), Dutch (NL), German (DE), Spanish (ES), Czech (CZ) and Polish (PL)) and there are as many files as classifications by medical specialties.

#### 3.1 Description of the XML tags

- **DisorderList count:** total number of clinical entities (disorders, group of disorders or subtypes of disorder) in the Xml file
- **Orphanum:** unique identifying number assigned by Orphanet to a given clinical entity
- **ExpertLink:** stable URL pointing to the specific page of the given clinical entity on the Orphanet website
- **Disorder\_Name:** preferred name of a given clinical entity
- **Lang:** ISO 639 code for language names
- **Name:** preferred name of a given clinical entity
- **ClassificationNodeList count:** number of clinical entities at the same level of the classification.
- **ClassificationNodeChildList count:** number of clinical entities under a given clinical entity

#### 3.2 Examples

```
<DisorderList count="10462">
```

10462 is the total number of clinical entities in this XML file

```
<OrphaNumber>68334</OrphaNumber>
```

```
<Name lang="en">Rare hemorrhagic disorder due to a coagulation factors defect</Name>
```

The concerned clinical entity has 68334 as its ORPHA number and "Rare hemorrhagic disorder due to a coagulation factors defect" as its preferred term

```
<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=68334</ExpertLink>
```

The stable URL pointing to information on this entry is [http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?lng=en&Expert=68334](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=68334)

```
<ClassificationNodeChildList count="2">
```

Two entries are present at this classification level.

```
<Orphanum>162949</Orphanum>
```

The classification ORPHA number is 162949.

```
<Name lang="en">Orphanet classification of rare hematological diseases</Name>
```

This is the name of the classification.

```
<ClassificationNodeChildList count="15">
```

There are 15 entries classified under the group of "Rare hemorrhagic disorder due to a constitutional coagulation factors defect".



## 4. Data subset relevant for data sharing at EU-level (Master file)

In the framework of the RD-Action Joint action (2015-2018), a file, namely the MasterFile, 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 including:

- Only the ORPHANumbers of disorder level in the Orphanet classification of Rare disorders (groups and subtypes of disorders are excluded);
- The preferred term and synonyms related to this ORPHANumbers subset;
- The cross referencing between these ORPHANumbers and the ICD-10 codes.

Offering an alignment of diagnostic terms with the ORPHANumber and ICD-10 code in a dataset, should minimize the bureaucratic burden of using different classifications and support standardization.

The content of the Masterfile can be easily extracted from the Orphanet nomenclature file and the Orphanet – ICD-10 cross referencing file:

- From the Orphanet nomenclature file, get only the Active ORPHANumbers at the Disorder level with their preferred term and synonyms;
- Then, from this data subset, query the Orphanet – ICD-10 cross referencing file to gather the cross referencing with the ICD-10 codes.

Example:

- a. In the Orphanet nomenclature file in English: in bold, the dataset relevant to retrieve for data sharing at EU-level.

```
</Disorder>
<Disorder id="99">
  <OrphaNumber>892</OrphaNumber>
  <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=892</ExpertLink>
  <Name lang="en">Von Hippel-Lindau disease</Name>
  The concerned clinical entity has 892 as its ORPHA number and Von Hippel-Lindau disease as its preferred term in English. Following
  http://www.orpha.net/consor/cgi-bin/OC\_Exp.php?lng=en&Expert=892 will open the page of the Orphanet website dedicated to
  the clinical entity.

  <FlagValue>1</FlagValue>
  <Totalstatus lang="en">Active</Totalstatus>
  The entity is in use in this version of the Orphanet Nomenclature.

  <SynonymList count="4">
    <Synonym lang="en">Familial cerebelloretinal angiomas</Synonym>
    <Synonym lang="en">Lindau disease</Synonym>
    <Synonym lang="en">VHL</Synonym>
    <Synonym lang="en">Von Hippel-Lindau syndrome</Synonym>
  </SynonymList>
  The entity has four synonyms in English.

  <GroupOfType id="36547">
    <Name lang="en">Disorder</Name>
  </GroupOfType>
  The entity is a disorder, not a group, not a subtype. Since it is active AND at the disorder level in the Orphanet classification, the
  ORPHANumber 892, its preferred term "Von Hippel-Lindau disease" and its four synonyms (Familial cerebelloretinal angiomas,
  Lindau disease, VHL and Von Hippel-Lindau syndrome) are to be used for data sharing at EU-level.

  <DisorderType id="21394">
    <Name lang="en">Disease</Name>
  </DisorderType>
  <DisorderDisorderAssociationList count="0">
  </DisorderDisorderAssociationList>
  <TextualInformationList count="1">
    <TextualInformation id="1260" lang="en">
```

```

<TextSectionList count="1">
  <TextSection id="50677" lang="en">
    <TextSectionType id="16907">
      <Name lang="en">Disease definition</Name>
    </TextSectionType>
    <Contents>Von Hippel-Lindau disease (VHL) is a familial cancer predisposition syndrome associated with a variety of malignant and benign neoplasms, most frequently retinal, cerebellar, and spinal hemangioblastoma, renal cell carcinoma (RCC), and pheochromocytoma.</Contents>
  </TextSection>
</TextSectionList>
</TextualInformation>
</TextualInformationList>
</Disorder>
<Disorder id="17569">
  <OrphaNumber>164823</OrphaNumber>
  <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=164823</ExpertLink>
  <Name lang="en">Rare acquired aplastic anemia</Name>
  The concerned clinical entity has 164823 as its ORPHA number and Rare acquired aplastic anemia as its preferred term in English.
  Following http://www.orpha.net/consor/cgi-bin/OC\_Exp.php?Ing=en&Expert=164823 will open the page of the Orphanet website dedicated to the clinical entity.

```

```

<FlagValue>513</FlagValue>
<Totalstatus lang="en">Active</Totalstatus>
The entity is in use in this version of the Orphanet Nomenclature.

```

```

<SynonymList count="0">
</SynonymList>

```

```

<GroupOfTypes id="36540">
  <Name lang="en">Group of disorders</Name>
</GroupOfTypes>

```

The entity is a group, not a disorder, not a subtype. Since it is active BUT NOT at the disorder level in the Orphanet classification, it should not be used to be used for data sharing at EU-level.

```

<DisorderType id="21436">
  <Name lang="en">Category</Name>
</DisorderType>
<DisorderDisorderAssociationList count="0">
</DisorderDisorderAssociationList>
<TextualInformationList count="0">
</TextualInformationList>
</Disorder>

```

- b. Once the relevant dataset from the Orphanet nomenclature file in English is retrieved, the Orphanet – ICD-10 cross referencing file is queried to get the cross referring to ICD-10:

```

<Disorder id="99">
  <OrphaNumber>892</OrphaNumber>
  <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Ing=en&Expert=892</ExpertLink>
  <Name lang="en">Von Hippel-Lindau disease</Name>
  <SynonymList count="4">
    <Synonym lang="en">Familial cerebelloretinal angiomatosis</Synonym>
    <Synonym lang="en">Lindau disease</Synonym>
    <Synonym lang="en">VHL</Synonym>
    <Synonym lang="en">Von Hippel-Lindau syndrome</Synonym>
  </SynonymList>

```

The entity has four synonyms in English. These synonyms are identical to the ones from the Orphanet nomenclature file. They can be retrieved similarly either from the Orphanet nomenclature file or from the Orphanet – ICD-10 cross referencing file.

```

<ExternalReferenceList count="1">
  <ExternalReference id="104856">
    <Source>ICD-10</Source>
    <Reference>Q85.8</Reference>
    <DisorderMappingRelation id="21534">
      <Name lang="en">NTBT (narrower term maps to a broader term)</Name>
    </DisorderMappingRelation>
    <DisorderMappingICDRelation id="21590">
      <Name lang="en">Inclusion term (The term is included under a ICD10 category and has not its own code)</Name>
    </DisorderMappingICDRelation>

```



```

<DisorderMappingValidationStatus id="21611">
  <Name lang="en">Validated</Name>
</DisorderMappingValidationStatus>
</ExternalReference>
</ExternalReferenceList>

```

This entity is mapped with ICD-10 reference "Q85.8". It is a narrower term that maps to the broader term of "Q85.8". The term is included in ICD-10 but under an ICD-10 category and does not have its own code. The relation between the reference and the clinical entity is "Validated". This cross referencing is intended to be used to support interoperability and standardization at the EU level.

```

</Disorder>

```

## 5. Appendix

**Definition:** Definition of the clinical entity.

**Disorder Mapping Relation:** Proximity of the relationship between an ORPHA clinical entity and the source entity. Can be either:

- an exact correspondence (E) when the concepts on both sides are equivalent;
- an asymmetric relationship (Orphanet concept broader or narrower than the concept in the target nomenclature): BTNT (the more generic term corresponds to one of the more specific terms) or NTBT (the more specific term corresponds to a more generic term);
- an incorrect match (W for Wrong, or two different concepts) but an exact syntactic match to a synonym or preferred term in the target terminology.

**DisorderMappingValidationStatus:** Validation status of the mapping between the ORPHA clinical entity and the reference. Can be either Validated or Not yet validated.

**ExpertLink:** stable URL pointing to the specific page of a given disease on the Orphanet website.

**Group of type:** Group of type of the clinical entity. Can be either (only one by clinical entity):

- Group of disorders: Clinical entity defined by a set of common features shared by several disorders and used to group them together. It can be a category or a clinical group.
- Disorder: Clinical entity defined by a set of phenotypic abnormalities with a homogeneous evolution and allowing for a definitive clinical diagnosis. It can be a disease, a malformation or clinical syndrome, a morphological or biological anomaly or a particular clinical situation in a disease or a syndrome.
- Subtype of disorder: Subdivision of a disorder. It can be a clinical subtype, an etiological subtype or a histopathological subtype.

**ID of the classification:** Unique, time-stable and non-reusable numerical identifier for the classification.

**Mapping ICD Relation:** Used only for ICD-10. Can be either:

- Specific code: the term of the Orphanet nomenclature has a specific code in ICD10;
- Inclusion term: the Orphanet nomenclature term corresponds to a term included in a code from the ICD10 tabular list, but does not have its own code;
- Index term: the term in the Orphanet nomenclature corresponds to a term present in the ICD10 index, and is absent from the tabular list but has a reference to a code in this list provided by the index;
- Attributed by Orphanet: the term of the Orphanet nomenclature is completely absent from the tabular list and index of the ICD10. An alignment is carried out by Orphanet according to established rules described in the document "[Orphanet\\_ICD10\\_coding\\_rules.pdf](#)".

**ORPHANumber:** Unique, time-stable and non-reusable numerical identifier. It is generated randomly by the database and named "ORPHACode" when used for coding purposes.

**ORPHANumberAggregation:** Recommended ORPHANumber in Europe for data sharing and statistical reporting. It encompasses the list of ORPHANumbers of Disorder typology, excluding groups and subtypes.

**Preferred term:** The most commonly accepted name in the medical community, according to published consensus, opinion of experts of the relevant medical specialty and/or compelling predominance of the name in medical literature. Preferred terms are unique throughout the database, associated to one ORPHANumber only.

**Synonym(s):** Terms that are perfectly equivalent to the preferred term. The number of synonyms is indefinite and may vary depending on the language of translation. Acronyms commonly used to describe the disease are included as synonyms.

**Total status:** Status of the clinical entity. Can be defined by only one status:

- Active: Active clinical entity in the nomenclature (in opposite to obsolete or deprecated entities that are no longer used in the nomenclature). The corresponding value can be 1, 513, 4097 or 4225.
- Inactive Deprecated entity: Inactive clinical entity formerly considered as distinct disorder and now recognised as being part of another disorder present in the database. The corresponding value is 257.
- Inactive Obsolete entity: Inactive clinical entity removed from the nomenclature for maintenance purposes. E.g. duplicate entities, categories not used anymore or non rare disorders. The corresponding value can be 16, 528 or 4112.
- Inactive Non Rare disease in Europe: Inactive clinical entity with a point prevalence in the general population less than 1/2'000. The corresponding value can be 48 or 1056.

**Type:** Type of the clinical entity. Can be either (only one by clinical entity):

- Biological anomaly: Disorder defined by a set of physiological abnormalities without clear associated clinical manifestations.
- Clinical subtype: Subdivision of a disorder further defined by its particular clinical presentation.
- Clinical syndrome: Disorder with a homogeneous evolution and homogeneous therapeutic possibilities, regardless of the physiopathological mechanism.
- Disease: Disorder with a homogeneous evolution and homogeneous therapeutic possibilities and a defined physiopathological mechanism. Excludes developmental anomalies.
- Etiological subtype: Subdivision of a disorder further defined by its aetiology.
- Clinical group: Group defined by a set of phenotypic abnormalities that are sufficient to establish a provisional diagnosis.
- Histopathological subtype: Subdivision of a disorder defined by the histological abnormalities in affected tissues.
- Malformation syndrome: Disorder resulting from a developmental anomaly involving more than one morphogenetic field. Includes sequences and associations.
- Morphological anomaly: Disorder defined by an alteration of the normal morphology resulting from a development anomaly involving a single morphogenetic field.
- Particular clinical situation in a disease or syndrome: Disorder occurring in particular circumstances.
- Category: Group with limited common features of which the use is limited to organising the Orphanet classifications.