orphanet

DESCRIPTION OF THE ORPHANET NOMENCLATURE FILES FOR CODING

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<!>**WARNING**<!>

The principal modifications made in this version are summarized here:

1) TotalStatus have **new FlagValue** from 2021
    - For “Inactive: Deprecated” entities: 8449 (formerly 257)
    - For “Inactive: Obsolete” entities: 8208, 9216 (formerly 16 and 1024, respectively)
    - For “Inactive: Non rare in Europe”: 8225 (formerly 48 or 1056)
2) Two classifications files have been removed from the Nomenclature pack from 2021:
    - ORPHAclassification_149_rare_sucking_swallowing_diseases_en.xml
    - ORPHAclassification_229_chromosomal_anomalies_sorted_by_chromosome_en.xml
3) Addition of a linearization file

<!>

The Orphanet nomenclature files for coding is a set of xml files that includes an Orphanet nomenclature file, an ICD-10 to Orphanet nomenclature mapping file, a linearization 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.

This pack, except for the linearization file which is only available in English, is available in 8 languages: Czech (CZ), Dutch (NL), English (EN), French (FR), German (DE), Italian (IT), Polish (PL), and Spanish (ES). This list may expand in the future.

The appropriate form when quoting Orphanet is: “*Orphanet: an online rare disease and orphan drug data base. © INSERM 1999. Available on http://www.orpha.net. Accessed (date accessed).*”

The appropriate form when quoting Orphadata is: “*Orphadata: Free access data from Orphanet. © INSERM 1999. Available on http://www.orphadata.org. Data version (XML data version).”*

1. Orphanet Nomenclature of Rare Diseases file

<!>WARNING<!>: TotalStatus have new FlagValue.

The Orphanet nomenclature is used to code the diagnosis of rare diseases with a specific identifier named the ORPHAcode in order to facilitate data collection, research and analysis. The Orphanet Nomenclature file contains all clinical 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 recommended by the European Joint Action RD-Action, a reduced list of ORPHAcodes has been agreed upon in order to allow data sharing and statistical analysis at EU-level. This list, based on the disorder level of the Orphanet classification, excluding groups and subtypes, establishes the “Aggregation level”.

For more information on definitions, please consult the Appendix of this document.

For more information on maintenance of the Orphanet nomenclature of rare diseases, please consult the procedure.

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.
- **ORPHAcode:** A unique and time-stable numerical identifier attributed randomly by the Orphanet database to each clinical entity upon its creation. Currently, the ORPHACode is made up of one to six digits. In the future, number of digits can expand. It comes with:
  - **ExpertLink:** stable URL pointing to the specific page of a given disease on the Orphanet website.
  - **Name:** the most generally accepted name for a clinical entity according to the literature, and as adopted by the medical community. Preferred terms are unique throughout the database, associated with one ORPHAcode only.
  - **Lang:** ISO 639 code for language name.
- **Totalstatus:** Status of the clinical entity. Can be either “Active”, “Inactive: Deprecated”, “Inactive: Obsolete”, “Inactive: Non rare disease in Europe” and only one by clinical entity.
- **Flagvalue:** Flag value of the clinical entity.
  For “Active” entities: 1, 129 or 513
  For “Inactive: Deprecated” entities: 8449 (formerly 257)
  For “Inactive: Obsolete” entities: 8208, 9216 (formerly 16 and 1024, respectively)
  For “Inactive: Non rare in Europe”: 8225 (formerly 48 or 1056)
- **Synonym(s):** A term that is perfectly equivalent to the preferred term of the clinical entity it is attached to. 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.
- **DisorderType:** An attribute used in the Orphanet database to characterise clinical entities according to their nosological definition within each level of classification. Can be either Category (id: 36561), Clinical group (id: 21436), Disease (id: 21394), Clinical syndrome (id: 21422), Malformation syndrome (id: 21401), Biological anomaly (id: 21408), Morphological anomaly (id: 21415), Particular clinical situation in a disease or syndrome (id: 21429), Etiological subtype (id: 21443), Clinical subtype (id: 21450) or Histopathological subtype (id: 21457) and only one by clinical entity.
- **ClassificationLevel:** A level of precision attributed to each clinical entity: Group of disorders (id: 36540), Disorder (id: 36547), or Subtype of disorder (id: 36554). These three levels organise the relational structure of the Orphanet classification.
- **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 “RootDisorder” and the Active entity for replacement as “TargetDisorder”.
- **SummaryInformation:** Textual information available for the ORPHAcode. Only definitions are provided here.
- **AggregationLevel:** Recommended ORPHAcode 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. To know more about the aggregation level, you can consult the document: **What is the Aggregation level?**

1.2 Examples

1.2.1 Active entity

Example 1: Disorder

```
<DisorderList count="10563">
10563 is the total number of clinical entities in this XML file

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

The concerned clinical entity has 5 as its ORPHAcode 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 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>
</SynonymList>
```

The entity has three synonyms in English.

```
<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.

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

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

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

As this entity is active, no association is provided.

```
<SummaryInformationList count="1">
 <SummaryInformation id="12552" lang="en">
  <TextSectionList count="1">
   <TextSection id="81974" lang="en">
    <TextSectionType id="16907">
     <Name lang="en">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>
 </SummaryInformation>
</SummaryInformationList>
```

A definition in English is available.

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

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

```
</Disorder>
```

Example 2: Group of disorders

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

The concerned clinical entity has 95498 as its ORPHAcode and Congenital anomaly of superior vena cava as its preferred term in English. Following http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=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>
</SynonymList>

The entity has two synonyms in English.

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

The entity is a category, not a clinical group.

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

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

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

As this entity is active, no association is provided.

<SummaryInformationList count="0">
</SummaryInformationList>

The definition is not available.

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

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

</Disorder>

Example 3: Subtype of disorders

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

The concerned clinical entity has 95626 as its ORPHAcode and Aduired central diabetes insipidus as its preferred term in English. Following 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>
</SynonymList>

The entity has two synonyms in English.

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

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

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

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

<DisorderDisorderAssociationList count="1">
<DisorderDisorderAssociation>
<TargetDisorder id="12650" cycle="true"/>

<RootDisorder id="12649">
<ORPHACode>95625</ORPHACode>
<Name lang="en">OBSOLETE: Posttraumatic diabetes insipidus</Name>
</RootDisorder>
<DisorderDisorderAssociationType id="27341">
<Name lang="en">Referred to</Name>
</DisorderDisorderAssociationType>
</DisorderDisorderAssociation>
</DisorderDisorderAssociationList>

The entity "OBSOLETE: Posttraumatic diabetes insipidus" has been obsoleted (RootDisorder). Acquired neurogenic diabetes insipidus should be used instead (TargetDisorder).

<SummaryInformationList count="1">
<SummaryInformation id="39282" lang="en">
<TextSectionList count="1">
<TextSection id="54203" lang="en">
<TextSectionType id="16907">
<Name lang="en">Definition</Name>
</TextSectionType>
<Contents>A subtype of central diabetes insipidus (CDI) characterized by polyuria and polydipsia, due to an idiopathic or secondary decrease in vasopressin (AVP) production.</Contents>
</TextSection>
</TextSectionList>
</SummaryInformation>
</SummaryInformationList>

A definition in English is available.

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

As this entity is a subtype, the ORPHAcode of the aggregation level is the closest disorder in the classification. Its ORPHAcode 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">
<ORPHACode>670</ORPHACode>
<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=670</ExpertLink>
<Name lang="en">PIBIDS syndrome</Name>

The concerned clinical entity has 670 as its ORPHAcode and PIBIDS as its preferred term in English. Following http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=670 will open the page of the Orphanet website dedicated to the clinical entity.

<FlagValue>8449</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>
</SynonymList>

The entity has two synonyms in English.

<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.

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

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

```
<DisorderDisorderAssociationList count="2">
```

There is 2 associations with this inactive entity. ORPHA:670 PIBIDS syndrome

```
 <DisorderDisorderAssociation>
  <TargetDisorder id="10319">
   <ORPHACode>33364</ORPHACode>
   <Name lang="en">Trichothiodystrophy</Name>
  </TargetDisorder>
  <RootDisorder id="963" cycle="true"/>
  <DisorderDisorderAssociationType id="21471">
   <Name lang="en">Moved to</Name>
  </DisorderDisorderAssociationType>
 </DisorderDisorderAssociation>
```

ORPHA:670 PIBIDS syndrome, identified as “RootDisorder”, is “moved to” the active entity ORPHA:33364 Trichothiodystrophy, identified as “TargetDisorder”.

```
 <DisorderDisorderAssociation>
  <TargetDisorder id="963" cycle="true"/>
  <RootDisorder id="1608">
   <ORPHACode>1408</ORPHACode>
   <Name lang="en">Hair defect-photosensitivity-intellectual disability syndrome</Name>
  </RootDisorder>
  <DisorderDisorderAssociationType id="21471">
   <Name lang="en">Moved to</Name>
  </DisorderDisorderAssociationType>
 </DisorderDisorderAssociation>
```

ORPHA:670 PIBIDS syndrome is also identified as “TargetDisorder” of another inactive entity ORPHA:1408 Hair defect-photosensitivity-intellectual disability syndrome.

```
</DisorderDisorderAssociationList>
```

Altogether, ORPHA:1408 Hair defect-photosensitivity-intellectual disability syndrome is “moved to” ORPHA:670 PIBIDS syndrome, itself moved to ORPHA:33364 Trichothiodystrophy.

```
<AggregationLevelSection>
 <AggregationLevelList count="1">
  <AggregationLevel>
   <ORPHACode>33364</ORPHACode>
   <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 ORPHAcode of replacement ORPHA:33364 Trichothiodystrophy is of disorder level, it can be used instead.

```
</Disorder>
```

Example 2: Obsolete entity

```
<Disorder id="8586">
 <ORPHACode>719</ORPHACode>
 <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&amp;Expert=719</ExpertLink>
 <Name lang="en">OBSOLETE: Pili canulati</Name>
```

The concerned clinical entity has 719 as its ORPHAcode and Pili canulati as its preferred term in English. Following http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=719 will open the page of the Orphanet website dedicated to the clinical entity.

```
 <FlagValue>8208</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">
</SynonymList>
The entity has 0 synonyms.

<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.

<ClassificationLevel id="36547">
<Name lang="en">Disorder<Name>
</ClassificationLevel>
The entity is a disorder, not a group, not a subtype.

<DisorderDisorderAssociationList count="1">
<DisorderDisorderAssociation>
<TargetDisorder id="1610">
<ORPHACode>1410</ORPHACode>
<Name lang="en">Uncombable hair syndrome</Name>
</TargetDisorder>
<RootDisorder 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 “RootDisorder”, is “referred to” the active entity ORPHA:1410 Uncombable hair syndrome, identified as “TargetDisorder”.

<AggregationLevelSection>
<AggregationLevelList count="1">
<AggregationLevel>
<ORPHACode>1410</ORPHACode>
<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 ORPHAcode 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">
<ORPHACode>1244</ORPHACode>
<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1244</ExpertLink>
<Name lang="en">NON RARE IN EUROPE: Bicuspid aortic valve</Name>
The concerned clinical entity has 1244 as its ORPHAcode and Bicuspid aortic valve as its preferred term in English. Following http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=1244 will open the page of the Orphanet website dedicated to the clinical entity.

<FlagValue>8225</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">
</SynonymList>
The entity has 0 synonyms.

<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.

<ClassificationLevel id="36547">
<Name lang="en">Disorder<Name>
</ClassificationLevel>
The entity is a disorder, not a group, not a subtype.

<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.

<SummaryInformationList count="1">
<SummaryInformation id="0.509" lang="en">
<TextSectionList count="0">
</TextSectionList>
<TextAuto>
<Info lang="en“> This disease is not rare in Europe. It does not belong to the Orphanet nomenclature of rare diseases.</Info>
</TextAuto>
</SummaryInformation>
</SummaryInformationList>
An automatic text stating that the disease is not rare in Europe is available in English.

<AggregationLevelSection>
<AggregationLevelList count="0"/>
<ORPHACodeAggregation/>
<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 ORPHAcode. Accordingly, no aggregation level is available.

</Disorder>

Example 4: Inactive entity doesn’t refer to a disorder

In some cases, the inactive entity is referred to a group or a subtype, since these levels are not used for data sharing and statistical reporting, the referred code cannot be used instead. Two solutions are possible:

- In case of referred group, the aggregation level is not applicable.
- In case of referred subtype, the aggregation level points the closest disorder out in the classification.

For more details, see the examples below.

4.1 Inactive entity referred to a group of disorders

<Disorder id="8773">
<ORPHACode>28455</ORPHACode>
<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=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 ORPHAcode 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?lng=en&Expert=28455 will open the page of the Orphanet website dedicated to the clinical entity.

<FlagValue>8208</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">
</SynonymList>
The entity has 0 synonyms.

<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.

<ClassificationLevel id="36547">
<Name lang="en">Disorder<Name>
</ClassificationLevel>
The entity is a disorder, not a group, not a subtype.

<DisorderDisorderAssociationList count="1">
<DisorderDisorderAssociation>
<TargetDisorder id="18292">
<ORPHACode>183625</ORPHACode>
<Name lang="en">Rare genetic diabetes mellitus</Name>
</DisorderOut>
<RootDisorder 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 “RootDisorder”, is “referred to” the active entity ORPHA:183625 Rare genetic diabetes mellitus, identified as “TargetDisorder”.

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

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

</Disorder>

4.2 Inactive entity referred to a subtype

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

The concerned clinical entity has 93609 as its ORPHAcode 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?lng=en&Expert=93609 will open the page of the Orphanet website dedicated to the clinical entity.

<FlagValue>8449</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.

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

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

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

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

<DisorderDisorderAssociationList count="1">
<DisorderDisorderAssociation>
<TargetDisorder id="22876">
<ORPHACode>402041</ORPHACode>
<Name lang="en">Autosomal recessive distal renal tubular acidosis</Name>
</TargetDisorder>
<RootDisorder id="12442" 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 "RootDisorder", is "moved to" the active entity ORPHA:402041 Autosomal recessive distal renal tubular acidosis, identified as "TargetDisorder".

```
  <AggregationLevelSection>
   <AggregationLevelList count="1">
    <AggregationLevel>
     <ORPHACode>18</ORPHACode>
     <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 ORPHAcode of replacement ORPHA:402041 Autosomal recessive distal renal tubular acidosis is of subtype level, it cannot be used instead. The closest ORPHAcode 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 ORPHAcode according to the Orphanet ICD-10 coding rules. The mapping includes a semantic link that specifies the relationship between an ORPHAcode 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
- **ORPHAcode:** A unique and time-stable numerical identifier attributed randomly by the Orphanet database to each clinical entity upon its creation. Currently, the ORPHACode is made up of one to six digits. In the future, number of digits can expand. It comes with:
  - **ExpertLink:** stable URL pointing to the specific page of a given disease on the Orphanet website.
  - **Name:** the most generally accepted name for a clinical entity according to the literature, and as adopted by the medical community. Preferred terms are unique throughout the database, associated with one ORPHAcode only.
  - **Lang:** ISO 639 code for language name.
- **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 ORPHAcode in the ICD-10
- **DisorderMappingRelation**: Closeness relationship between an ORPHAcode 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 ORPHAcode 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 ORPHAcode 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: ORPHAcode has a specific code in ICD10

```
<Disorder id="109">
    <ORPHACode>558</ORPHACode>
    <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&amp;Expert=558</ExpertLink>
    <Name lang="en">Marfan syndrome</Name>
```

The concerned clinical entity has 558 as its ORPHAcode and Marfan syndrome as its preferred term in English. Following http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=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 two concepts are equivalent)</Name>
        </DisorderMappingRelation>
        <DisorderMappingICDRelation id="21583">
          <Name lang="en">Specific code (The ORPHA code 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: ORPHAcode has an inclusion term on ICD10

```
<Disorder id="106">
    <ORPHACode>803</ORPHACode>
    <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&amp;Expert=803</ExpertLink>
    <Name lang="en">Amyotrophic lateral sclerosis</Name>
```

The concerned clinical entity has 803 as its ORPHAcode and Amyotrophic lateral sclerosis as preferred term in English. Following http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=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 (ORPHA code's Narrower Term maps to a Broader Term)</Name>
        </DisorderMappingRelation>
        <DisorderMappingICDRelation id="21590">
          <Name lang="en">Inclusion term (The ORPHA code 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: ORPHACode has an index term on ICD10

```
<Disorder id="548">
  <ORPHACode>635</ORPHACode>
  <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&amp;Expert=635</ExpertLink>
  <Name lang="en">Neuroblastoma</Name>
```
The concerned clinical entity has 635 as its ORPHAcode and Neuroblastoma as its preferred term in English. Following http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=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 (ORPHA code's Narrower Term maps to a Broader Term )</Name>
      </DisorderMappingRelation>
      <DisorderMappingICDRelation id="21597">
        <Name lang="en">Index term (The ORPHA code is listed in the ICD10 Index)</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: ORPHACode has an ICD10 term attributed by Orphanet

```
</Disorder>
  <Disorder id="553">
    <ORPHACode>2746</ORPHACode>
    <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&amp;Expert=2746</ExpertLink>
    <Name lang="en">Opsismodysplasia</Name>
```
The concerned clinical entity has 2746 as its ORPHAcode and Opsismodysplasia as itspreferred term in English. Following http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=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 (ORPHA code's Narrower Term maps to a Broader Term)</Name>
        </DisorderMappingRelation>
        <DisorderMappingICDRelation id="21604">
          <Name lang="en">Attributed (The ICD10 code is attributed by Orphanet)</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

<!>WARNING<!>

Two classifications files have been removed from the Nomenclature pack from 2021:

- ORPHAclassification_149_rare_sucking_swallowing_diseases_en.xml
- ORPHAclassification_229_chromosomal_anomalies_sorted_by_chromosome_en.xml

<!>

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.

4.1 Description of the XML tags

- **ClassificationList count:** total number of classification in the Xml file. Usually only 1.
- **OrphaNumber:** unique and time-stable numerical identifier attributed randomly by the database.
- **ClassificationNodeRootList count:** number of clinical entities at upper level of the hierarchy (number of clinical entities without parent). Usually only 1.
- **ClassificationNode**: level in the classification where a clinical entity has at least one parent. It may have child or not.
- **ClassificationNodeChildList count:** number of clinical entities under a given clinical entity.
- **ORPHAcode:** A unique and time-stable numerical identifier attributed randomly by the Orphanet database to each clinical entity upon its creation. Currently, the ORPHACode is made up of one to six digits. In the future, number of digits can expand. It comes with:
    - **ExpertLink:** stable URL pointing to the specific page of a given disease on the Orphanet website.
    - **Name:** the most generally accepted name for a clinical entity according to the literature, and as adopted by the medical community. Preferred terms are unique throughout the database, associated with one ORPHAcode only.
    - **Lang:** ISO 639 code for language name.

4.2 Examples

```
<ClassificationList count="1">
  <Classification id="146">
    <OrphaNumber>156265</OrphaNumber>
    <Name lang="en">Orphanet classification of rare cardiac diseases</Name>
```
This Xml file includes 1 classification. Its unique Identifier is 156265 and it is named Orphanet classification of rare cardiac diseases.

```
    <ClassificationNodeRootList count="1">
```

This classification has only one root.

<ClassificationNode>

The following clinical entity is a node in the classification: it has at least one parent in the classification.

<Disorder id="18899">
<ORPHACode>218439</ORPHACode>
<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=218439</ExpertLink>
<Name lang="en">Non-genetic cardiac rhythm disease</Name>

This clinical entity has 218439 as its ORPHAcode and "Non-genetic cardiac rhythm disease" as its preferred term. The stable URL pointing to information on this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=218439.

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

It is a category.

<ClassificationNodeChildList count="3">

The node ORPHA:218439 "Non-genetic cardiac rhythm disease" has three children.

<ClassificationNode>

The first child is a node in the classification: it has at least one parent.

<Disorder id="8617">
<ORPHACode>3282</ORPHACode>
<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3282</ExpertLink>
<Name lang="en">Multifocal atrial tachycardia</Name>
<DisorderType id="21394">
<Name lang="en">Disease</Name>
</DisorderType>
</Disorder>

The first child has 3282 as its ORPHAcode and "Multifocal atrial tachycardia" as its preferred term. The stable URL pointing to information on this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=3282.It is a disease.

<ClassificationNodeChildList count="0">
</ClassificationNodeChildList>

ORPHA:3282 "Multifocal atrial tachycardia" doesn't have child.

<ClassificationNode>

The second child is a node in the classification: it has at least one parent.

<Disorder id="10590">
<ORPHACode>45452</ORPHACode>
<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=45452</ExpertLink>
<Name lang="en">Idiopathic neonatal atrial flutter</Name>
<DisorderType id="21394">
<Name lang="en">Disease</Name>
</DisorderType>
</Disorder>

The second child has 45452 as its ORPHAcode and "Idiopathic neonatal atrial flutter" as its preferred term. The stable URL pointing to information on this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=45452.It is a disease.

<ClassificationNodeChildList count="0">
</ClassificationNodeChildList>

ORPHA:45452 "Idiopathic neonatal atrial flutter" doesn't have child.

<ClassificationNode>

The third and last child is a node in the classification: it has at least one parent.

<Disorder id="10591">
<ORPHACode>45453</ORPHACode>
<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=45453</ExpertLink>
<Name lang="en">Incessant infant ventricular tachycardia</Name>
<DisorderType id="21394">
<Name lang="en">Disease</Name>
</DisorderType>
</Disorder>

The third child has 45453 as its ORPHAcode and "Incessant infant ventricular tachycardia" as its preferred term. The stable URL pointing to information on this entry is http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=45453.It is a disease.

<ClassificationNodeChildList count="0">
</ClassificationNodeChildList>
ORPHA:45453 “Incessant infant ventricular tachycardia” doesn’t have child.

</ClassificationNode>

4. Orphanet Linearisation file

<!>WARNING<!>: This is a new data set file added in 2021 to the Nomenclature pack.

The linearization is a process applied in the Orphanet database to attribute one head of classification (called Preferential parent) to each clinical entity, in order to enable the sorting out of all clinical entities by medical specialty and avoid multiple counting of multiclassified entities in statistical analysis. As some decisions could be made somewhat arbitrarily, we have written a set of rules to make sure attributions are consistent. The methodology can be found here.

4.1 Description of the XML tags

- **DisorderList count**: total number of clinical entities (disorders, group of disorders or subtypes of disorder) in the Xml file.
- **ORPHAcode:** A unique and time-stable numerical identifier attributed randomly by the Orphanet database to each clinical entity upon its creation. Currently, the ORPHACode is made up of one to six digits. In the future, number of digits can expand. It comes with:
  - **ExpertLink:** stable URL pointing to the specific page of a given disease on the Orphanet website.
  - **Name:** the most generally accepted name for a clinical entity according to the literature, and as adopted by the medical community. Preferred terms are unique throughout the database, associated with one ORPHAcode only.
  - **Lang:** ISO 639 code for language name.
- **DisorderDisorderAssociation:** Relationship between a clinical entity and one head of Orphanet classification.

4.2 Example :

<DisorderList count="7513">
7513 is the total number of clinical entities in this XML file

<Disorder id="17601">
<OrphaCode>166024</OrphaCode>
<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=166024</ExpertLink>
<Name lang="en">Multiple epiphyseal dysplasia, Al-Gazali type </Name>
The concerned clinical entity has 166024 as its ORPHAcode and Multiple epiphyseal dysplasia, Al-Gazali type as preferred term in English. Following http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=166024 will open the page of the Orphanet website dedicated to the clinical entity..

<DisorderDisorderAssociationList count="1">
<DisorderDisorderAssociation>
<TargetDisorder id="12333">
<OrphaCode>93419</OrphaCode>
<Name lang="en">Rare bone disease </Name>
</TargetDisorder>
<RootDisorder id="17601" cycle="true">
<DisorderDisorderAssociationType id="21485">
<Name lang="en">Preferential parent</Name>
</DisorderDisorderAssociationType>
</DisorderDisorderAssociation>
</DisorderDisorderAssociationList>
This entity has the group ORPHA: 93419 Rare bone disease as preferential parent

</Disorder>

5. 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 ORPHAcodes 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 ORPHAcodes subset;
- The cross referencing between these ORPHAcodes and the ICD-10 codes.

From 2021, the Master file is provided in the nomenclature pack in a single English version and in xlsx format. However, you can easily produce it in your national language by using the Orphanet nomenclature file and the Orphanet - ICD-10 cross referencing file and following the steps below:

- From the Orphanet nomenclature file of your national language, get only the Active ORPHAcodes at the Disorder level (FlagValue: 1, 129 or 513 and ClassificationLevel id="36547") 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">
    <ORPHACode>892</ORPHACode>
    <ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&amp;Expert=892</ExpertLink>
    <Name lang="en">Von Hippel-Lindau disease</Name>
```

The concerned clinical entity has 892 as its ORPHAcode 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 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.

```
    <DisorderType id="21394">
      <Name lang="en">Disease</Name>
    </DisorderType>
    <ClassificationLevel id="36547">
      <Name lang="en">Disorder<Name>
    </ClassificationLevel>
```

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 ORPHAcode 892, its preferred term “Von Hippel-Lindau disease” and its four synonyms (Familial cerebelloretinal angiomatosis, Lindau disease, VHL and Von Hippel-Lindau syndrome) are to be used for data sharing at EU-level.

```
    <DisorderDisorderAssociationList count="0">
    </DisorderDisorderAssociationList>
    <SummaryInformationList count="1">
```

<SummaryInformation id="1260" lang="en">
<TextSectionList count="1">
<TextSection id="50677" lang="en">
<TextSectionType id="16907">
<Name lang="en">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>
</SummaryInformation>
</SummaryInformationList>
</Disorder>
<Disorder id="17569">
<ORPHACode>164823</ORPHACode>
<ExpertLink lang="en">http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=en&Expert=164823</ExpertLink>
<Name lang="en">Rare acquired aplastic anemia</Name>

The concerned clinical entity has 164823 as its ORPHAcode and Rare acquired aplastic anemia as its preferred term in English. Following http://www.orpha.net/consor/cgi-bin/OC_Exp.php?lng=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>

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

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="36561">
<Name lang="en">Category</Name>
</DisorderType>
<DisorderDisorderAssociationList count="0">
</DisorderDisorderAssociationList>
<SummaryInformationList count="0">
</SummaryInformationList>
</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">
<ORPHACode>892</ORPHACode>
<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>
<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 (ORPHA code's Narrower Term maps to a Broader Term)</Name>
</DisorderMappingRelation>
<DisorderMappingICDRelation id="21590">
<Name lang="en">Inclusion term (The ORPHA code 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>
```


6. Appendix

Definition: A short text stating the group of disorders that the clinical entity belongs to, and listing the major clinical characteristics (e.g. clinical, pathological, radiological, etc.) that define the entity and differentiate it from other entities classified within the same clinical group.

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.

Classification level: A level of precision attributed to each clinical entity: Group of disorders, Disorder, or Subtype of disorder. These three levels organise the relational structure of the Orphanet classification:

- Group of disorders (id: 36540): a collection of clinical entities sharing a set of common features. It can be a category or a clinical group.
- Disorder (id: 36547): a clinical entity characterised by a set of homogeneous phenotypic abnormalities and evolution allowing a definitive clinical diagnosis.
- Subtype of disorder (id: 36554): a subdivision of a disorder according to a positive criterion.

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".

ORPHAcode: a unique and time-stable numerical identifier attributed randomly by the Orphanet database to each clinical entity upon its creation. Currently, the ORPHAcode is made up of one to six digits. In the future, number of digits can expand.

ORPHACodeAgregation: Recommended ORPHAcode in Europe for data sharing and statistical reporting. It encompasses the list of ORPHAcodes of Disorder typology, excluding groups and subtypes.

Preferred term: The most generally accepted name for a clinical entity according to the literature, and as adopted by the medical community.

Synonym: a term that is perfectly equivalent to the preferred term of the clinical entity it is attached to. As many synonyms as necessary are added to a preferred term. Acronyms are included only when they are consistently used in the literature. .

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

- Active: A clinical entity that is in use in the nomenclature (in opposite to obsolete or deprecated entities that are no longer used in the nomenclature). The corresponding value can be 1, 129 or 513.
- Inactive: A clinical entity that has been excluded from the Orphanet nomenclature. This includes obsolete entities, deprecated entities, and entities that have been inactivated because they are not rare in Europe :
    - Inactive: A clinical entity that was initially considered as an independent diagnosis, but is now considered as part of another diagnosis as a result of the evolution of knowledge, and is therefore removed from the Orphanet nomenclature. The corresponding value is 8449.
    - Inactive: Obsolete entity: A clinical entity that has been removed from the Orphanet nomenclature for one of the following reasons: exact duplicate of another existing clinical entity; unclear clinical entity that cannot be precisely characterised; only one published case in the literature; organisational category that is no longer in use. The corresponding value is 8208 or 9216.
    - Inactive: Non Rare disease in Europe: A clinical entity that does not meet the European definition of a rare disease (less than 5 affected individuals per 10,000 in Europe) in light of current epidemological knowledge, and has therefore been removed from the Orphanet nomenclature. The corresponding value is 8225.

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

- Biological anomaly (id: 21408): Disorder defined by a set of physiological abnormalities without clear associated clinical manifestations.
- Clinical group (id: 21436): A group of clinically homogeneous disorders that share a similar etiology, course, outcome, and/or management.
- Clinical subtype (id: 21450): Subdivision of a disorder according to distinct clinical characteristics (severity, age of onset, particular clinical signs, etc.).
- Clinical syndrome (id: 21422): A disorder with homogeneous therapeutic possibilities, regardless of the pathophysiological mechanism involved.
- Disease (id: 21394): A disorder with homogeneous therapeutic possibilities and an identified pathophysiological mechanism. Developmental anomalies are excluded.
- Etiological subtype (id: 21443): Subdivision of a disorder according to distinct causes resulting in a similar clinical presentation.
- Histopathological subtype (id: 21457): Subdivision of a disorder according to characteristic histological patterns.
- Malformation syndrome (id: 21401): A disorder resulting from a developmental anomaly involving more than one morphogenetic field. Malformative sequences and associations are included.
- Morphological anomaly (id: 21415): A disorder characterised by a morphological alteration resulting from a development anomaly involving a single morphogenetic field.

- Particular clinical situation in a disease or syndrome (id: 21429): A set of phenotypic abnormalities presenting in a subset of patients under particular circumstances.
- Category (id: 36561): A group of clinically heterogeneous disorders sharing one general feature, used to organise the classification.