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SNOMED CT and Orphanet – working together

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Delivering SNOMED CT The global language of healthcare

Session outline

- What is Orphanet?
- Rare disorders
- Orphanet nomenclature
- Mappings to other terminologies
- IHTSDO/INSERM agreement
- Use cases
- Benefits
- SNOMED CT content changes
- Gap analysis
- Timescales
- Future direction

Orphanet nomenclature of rare diseases and their representation in SNOMED CT

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What is Orphanet nomenclature?

- Orphanet is the **leader in collecting, integrating, producing and dissemination of added-value information and data in the field of rare diseases**.
- Orphanet is the reference for the **nomenclature and classification of rare diseases** (the only specific resource) and has a normative role in this domain.
- **Orphanet nomenclature is produced by the INSERM in France**
- **The European Commission expert Group for rare Diseases adopted a recommendation for EU member states to introduce ORPHA codification in health inforamtion systems (November 2014)**

Rare disorders in Orphanet

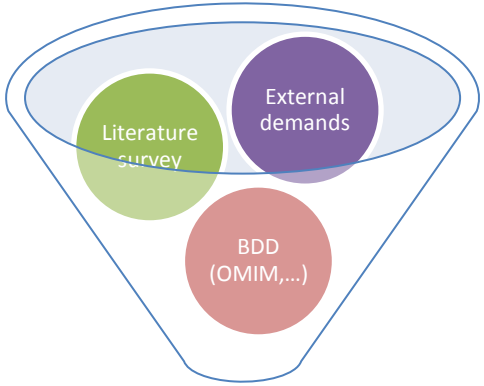
- Since 1997: **Inventory** of rare disorders
  - (prevalence <1/2 000)
  - Alignements to OMIM
- 2005: alignments to ICD-10
- 2007: **Classification** of rare disorders
- 2011: Alignments to UMLS, MeSH, MedDRA, and to SNOMED CT through UMLS
- 2014: ORDO (Orphanet **ontology** of rare diseases) in collaboration with the EBI (Hinxton, UK).

Orphanet nomenclature

- Each entry (group, disorder, subtype) is given a unique, stable **ORPHA number**
  - The Orphacode
- Each entry is given a preferred term and as many synonyms as necessary
- Nomenclature is translated (FR – ES – IT – PT – DE – NL)
- Nomenclature and classifications are updated monthly.

Orphanet nomenclature

- Produced by the INSERM (France), hosting Orphanet, the EU RD database
  - Institutional support
- Under CreativeCommons
  - Free for re-use
- Strong position in international terminologies
  - Ongoing process to be a WHO collaborating center
    - Population and updates of ICD11: TAG-RD leaders
  - Ongoing MoU with IHTSDO (SNOMED CT)
    - Population of SNOMED CT and mappings



Nomenclature update process

Verification: Orphanet, literature, expert advice

Qualifying the demand

Verification: prevalence, needs for representation (i.e. orphan drug)

Creation

Modification

Decision

Diseases committee

+ info

Rejection

New entry

Nomenclature

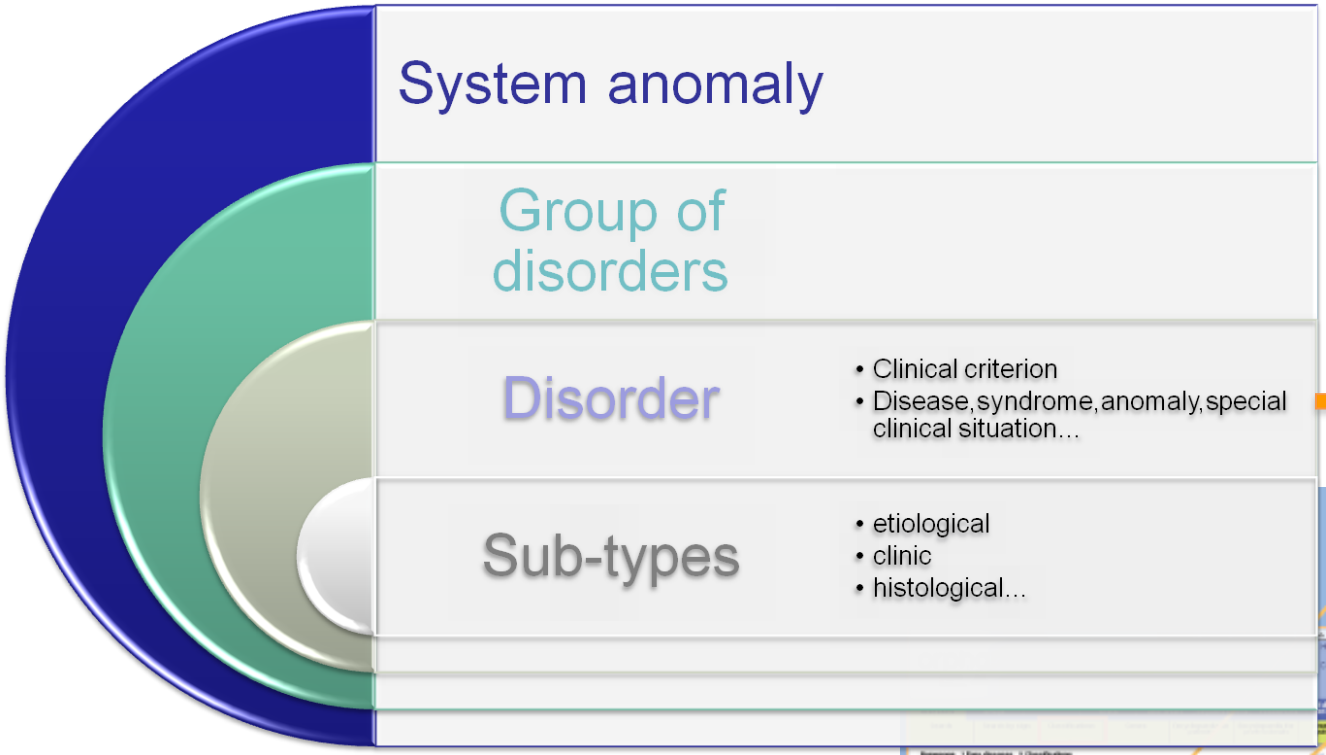
Obsolescence

Deprecation

Expert advice

Impact on classifications:
New entries,
inter- and intra-classification consistency
Creation of organising categories

A multidimensional classification



5,873 rare disorders

Rare metabolic disease
Metabolic disease involving complex molecules
Peroxisomal disease
Adrenoleukodystrophy, X-linked
Adrenoleukodystrophy, X-linked, cerebral form
Adrenomyeloneuropathy

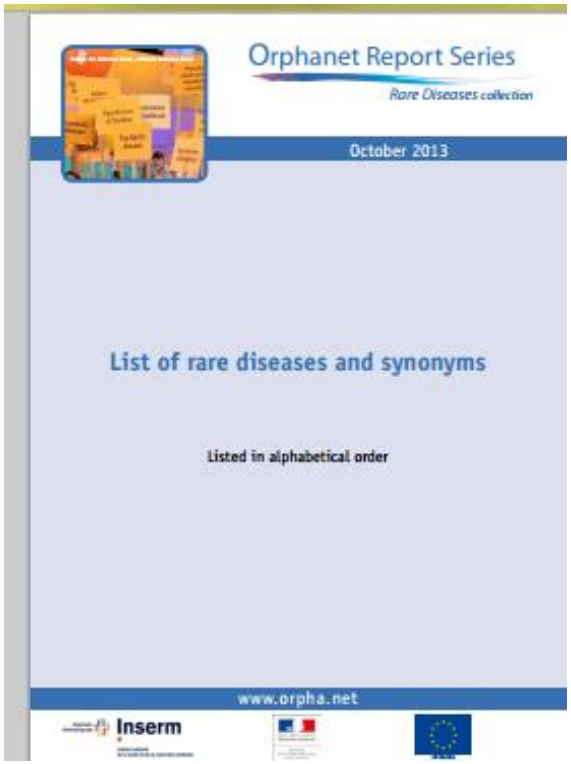
Rare neurologic disease
Neurometabolic disease
Adrenoleukodystrophy, X-linked
Adrenoleukodystrophy, X-linked, cerebral form
Adrenomyeloneuropathy

Rare neurologic disease
Rare epilepsy
Metabolic diseases with epilepsy
Peroxisomal disease
Adrenoleukodystrophy, X-linked
Adrenoleukodystrophy, X-linked, cerebral form
Adrenomyeloneuropathy

Rare neurologic disease
Leukodystrophy
Adrenoleukodystrophy, X-linked
Adrenoleukodystrophy, X-linked, cerebral form
Adrenomyeloneuropathy

Rare endocrine disease
Rare adrenal disease
Primary adrenal insufficiency
Chronic primary adrenal insufficiency
Genetic chronic primary adrenal insufficiency
Adrenoleukodystrophy, X-linked
Adrenoleukodystrophy, X-linked, cerebral form

To find the Orphanet nomenclature



Alphabetical list of names and synonyms
with ORPHA code
Updated every 6 months
Translated.
PDF (download, print)

ORPHA nomenclature
Cross-references
Monthly updated
6 languages (7 soon)
For download
XML

Free access data from Orphanet
orphanet

orphadata

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Freely accessible datasets

Diseases, cross referenced with other nomenclatures

February-2014

Rare Diseases And Cross-Referencing

Files available in XML format.

Rare diseases and cross-referencing

Language	links	size
English	http://www.orphadata.org/data/xml/en_product1.xml	6.14 MB
French	http://www.orphadata.org/data/xml/fr_product1.xml	6.15 MB
Spanish	http://www.orphadata.org/data/xml/es_product1.xml	6.10 MB
Italian	http://www.orphadata.org/data/xml/it_product1.xml	6.05 MB
Portuguese	http://www.orphadata.org/data/xml/pt_product1.xml	5.81 MB
German	http://www.orphadata.org/data/xml/de_product1.xml	5.99 MB

Mappings to other terminologies

- Disorders mapped to OMIM (manually)
- Disorders mapped to ICD-10 (manually)
- Disorders mapped to UMLS, MeSH, MedDRA (semi-automatically; manually curated)
- **Mappings are qualified** (exact ; narrow-to-broad ; broad-to-narrow)
- Information on the validation status is noted
- Updates depending on the target terminology
  - Monthly (ICD10, OMIM)
  - Twice a year (UMLS, MeSH, MedDRA)

Qualifying mappings

E	exact mapping (the terms and the concepts are equivalent)
NTBT	narrower term maps to a broader term
BTNT	broader term maps to a narrower term
W	incorrect mapping (two different concepts)
NTBT/E	narrower term maps to a broader term because of an exact mapping with a synonym in the target terminology
BTNT/E	broader term maps to a narrower term because of an exact mapping with a synonym in the target terminology
W/E	incorrect mapping (two different concepts) but syntactically exact mapping to a synonym or a preferred term in the target terminology
ND	not yet decided/unable to decide
The following are attributed to ICD10 codes only :	
Specific code	The term has its own code in the ICD10
Inclusion term	The term is included under a ICD10 category and has not its own code
Index term	The term is oncluded in ICD10 index and refers to one more general code
Attributed code	The term does not exist in ICD10 and a code was attributed by Orphanet

Details of the Agreement

- IHTSDO and INSERM * signed Collaboration Agreement in May 2015
- Focus of the agreement is Orphanet (inventory & classification of rare diseases)
- Creation of joint works beneficial to both organizations
- In the interest of both parties to collaborate
- Supports EPR/EHR and secondary uses

* INSERM - Institut national de la santé et de la recherche médicale - Owners of Orphanet

Scope of the Agreement

- Focus of the Agreement is Orphanet, to provide:
  - Additional Rare Disease content for SNOMED CT
  - Preferred terms/synonyms for rare disease content
  - Text definitions for rare disease content
- To provide a complete linkage between SNOMED CT and Orphanet
- Other products owned by INSERM are currently out of scope

Use cases/benefits for users

- EHR recording of rare diseases
  - Recording of diagnosis in the context of the patient record
- Central reporting of rare disease data
- Secondary analysis of rare disease data
- Analysis of rare disease data based on gene mutation

Benefits to SNOMED CT

- Increased coverage of rare diseases in SNOMED CT International release
- Provides validated text definitions for rare disease content
- Provides the foundation for future work on genetics/genomics
- Supports secondary usage of data through provision of linkage table

Changes to SNOMED CT content

- SNOMED CT has rare disease coverage, but is incomplete
- Content contained under Disorders
- Orphanet provides
  - Increased coverage
  - Preferred terms
  - Synonyms
  - Text definitions

Orphanet and SNOMED CT

- Workplan so far:
  - Produce candidate matching terms between Orphanet nomenclature (all semantic types) and SNOMED CT (semantic types: disease, morphological abnormality, finding)
  - Expert review
    - The relationship between terms are qualified according to the definitions presented
  - Gap analysis:
    - List of rare disorders lacking in SNOMED CT (PT & synonyms)
  - Incorporate lacking terms (rare disorders) to SNOMED CT
  - Produce a mapping file for interoperability

SNOMED CT July 2015 International release

Gap analysis

- 6320 alignments were proposed for 5191 Orphanet entries
  - 3203 unique Orpha entries have at least one EXACT match in SNOMED
- After exclusion of semantic types: group, subtype, obsolete entry, deprecated entry:
  - 2343/5873 ORPHA disorders are included in SNOMED CT (39.89%)
- GAP: 3530 ORPHA disorders (excluding groups and subtypes)
  - 12,848 terms when PT, synonyms and acronyms are included

Timescales

- June – July 2015
  - Creation of default matching table, using an automated algorithmic mapping approach
- August 2015
  - Delivery of list of concept additions required for addition to the SNOMED CT International Release
- January – April 2016
  - Authoring of new content and authoring of any required changes to SNOMED CT by IHTSDO (editing team)
- May – June 2016
  - Creation of SNOMED CT to Orphanet mapping table
- July 2015
  - Release of new content in SNOMED CT

. . . the future . . .

- Future development of SNOMED CT content to support genetics/genomics
- Leverage additional INSERM resources to support genetics/genomics subject to future requirements
  Explore development of additional products based Orphanet content

Thank you for listening

Merci de votre écoute

For more information:

http://www.orpha.net
http://www.ihtsdo.org