Orphanet and ICD-11
Tackling the Rare Diseases codification challenge

Ana Rath
INSERM, US14 – Orphanet
ana.rath@inserm.fr
### Challenges in RD data

<table>
<thead>
<tr>
<th>Challenge</th>
<th>Detail</th>
</tr>
</thead>
<tbody>
<tr>
<td>Rarity!</td>
<td>International is the right scale</td>
</tr>
<tr>
<td>Domain evolutivity</td>
<td>New RD are frequently described</td>
</tr>
<tr>
<td>Heterogeneity</td>
<td>Need for cross-domain harmonisation</td>
</tr>
<tr>
<td>Portability</td>
<td>For a consistent healthcare pathway</td>
</tr>
<tr>
<td>Reusability</td>
<td>For knowledge generation based on data</td>
</tr>
<tr>
<td>Invisibility</td>
<td>RD are ill-represented in health terminologies: need for specific codification</td>
</tr>
</tbody>
</table>
Representing rare diseases: ORPHA CODES

A MEDICAL TERMINOLOGY SPECIFIC TO RARE DISEASES (<1 in 2000 cases)

Clinical definition:
Disorders are clinically homogeneous entities described in at least two independent individuals, confirming that the clinical signs are not associated by fortuity.

Group

Disorder

Subtype

"Classification level"

9,339 clinical entities
2,097 groups
6,227 disorders
1,014 subtypes

Comprehensive, standardized, evidence-based, interoperable, versioned, computable and free (CC-BY 4.0)
Orphanet and WHO: a long-lasting collaboration

- ORPHAcodes-ICD-10 mappings curated and distributed by Orphanet
- ICD-11
  - Orphanet chaired the Rare Diseases -Topic Advisory Group (RD-TAG)
  - Orphanet took part in the ICD-11 Revision Steering Committee
  - Orphanet contributes to populate ICD-11 foundation with
    - Nomenclature (terms)
    - Definitions
- Orphanet France (INSERM) is part of the French WHO-FIC Collaborating Centre
  - Lead by Agence du Numérique en Santé (ANS)
Our common goal

- Increase RD visibility in health records
  - To generate data for primary and secondary use
- Allow for cross-border accurate and reliable RD reporting
  - To improve epidemiological knowledge on RD
- Allow for cross-sector interoperability
  - e.g. Between EHRs and registries not using ICD
- Facilitate the implementation of ORPHAcodes together with ICD-11 in health information systems
  - To facilitate RD coding and transcoding with ICD-11
- Improve the ICD-11 content
  - By interconnecting Orphanet knowledge base diversity of data
Towards general interoperability for RD

http://id.who.int/icd/entity/426701963
Next steps

- Producing a curated mapping file (WIP)
  - Including the characterisation of the relation between codes
    - Exact or not
    - If not exact, how

- Increasing Rare Diseases content in ICD-11
  - Orphanet nomenclature relies on
    - A worldwide network of experts
    - Published methodologies*

- Contribute to genomic information, definitions and other scientific content to ICD-11

* [http://www.orpha.net/ophacom/cahiers/docs/GB/eproc_Disease_naming_rules_in_English_PR_R1_Nom_01.pdf](http://www.orpha.net/ophacom/cahiers/docs/GB/eproc_Disease_naming_rules_in_English_PR_R1_Nom_01.pdf)
In conclusion

- Will allow for a better representation of Rare Diseases in health information systems
- Orphanet and WHO are exploring ways to synergise in the future
  - In order to increase the uptake of a specific Rare Diseases codification
  - Based on the uniqueness of Orphanet methodology for maintenance of a specific Rare Diseases nomenclature as knowledge evolves
  - Based on the expertise, technology and worldwide outreach of WHO
Thank you for your attention

www.orpha.net
www.orphadata.com