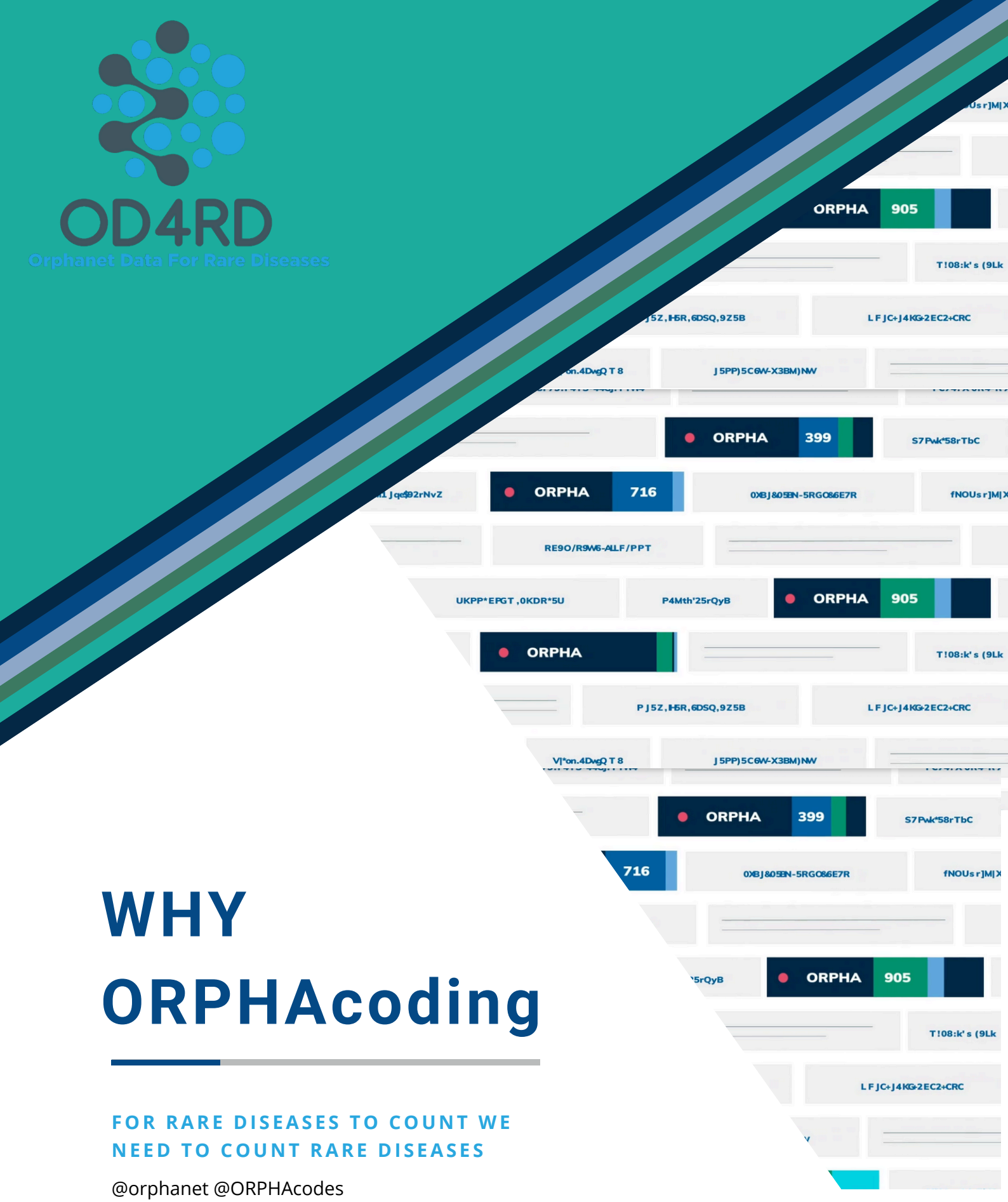
OD4RD

Orphanet Data For Rare Diseases



WHY ORPHAcoding

FOR RARE DISEASES TO COUNT WE NEED TO COUNT RARE DISEASES

@orphanet @ORPHAcodes

ORPHA CODES

Co-funded by the European Union

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RARE DISEASES ARE NOT VISIBLE WHEN USING GENERIC TERMINOLOGIES ONLY

Underrepresented

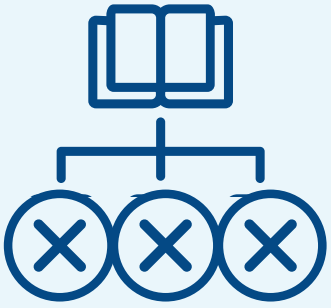
ICD-10: ~83% of RD do not have any code*
ICD-11: ~37% of RD do not have any code*
SNOMED CT: 7% of RD do not have any code*
None of these terminologies have a code for the patients in a diagnostic dead-end/undiagnosed RD patients

Imprecise

ICD-10: 93 % of RD do not have a PRECISE code*
ICD-11: 75 % of RD do not have a PRECISE code*
SNOMED CT: 7 % of RD do not have a PRECISE code*

Not classified as “rare”

RD are "lost" amongst common diseases
Generic terminologies are not exploitable for RD-specific statistics



*All figures presented are exact as of 01 September 2023.

UNMET RARE DISEASE PATIENTS' NEEDS & INEFFICIENT HEALTHCARE PLANNING

Not all RD diagnoses are traceable

- **Inadequate health planning** for RD diagnoses and undiagnosed patients that cannot be represented by generic terminologies

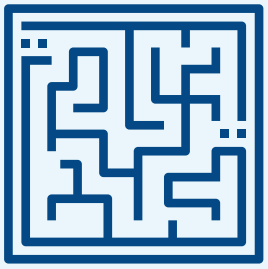
RD diagnoses cannot be traced precisely

- **Impaired capacity of Healthcare Systems to address the SPECIFIC NEEDS** of these patients, many of which are **vital**

Lost amongst common diseases

- **Limited healthcare monitoring** in regard to a given RD or a group of RD, or RD as a whole.
- **Difficult & time consuming** to produce **RD indicators**

including ERNs' evaluations or contributions to epidemiological knowledge....



ORPHACODES MAKE RARE DISEASES VISIBLE

... alongside the generic terminology in use

100% of RD represented

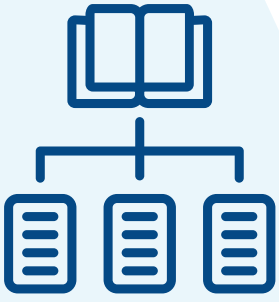
- **ALL RD diagnoses** are represented in ORPHAcodes including the undiagnosed
- They provide **accurate representation** of the current knowledge and meet coders' needs*

Precise

ORPHAcodes include each RD, including subtypes of diseases; this will ensure that **ALL PATIENT NEEDS are met** including the very specific ones

Classified

The specific RD-specific classification system allows **effective healthcare monitoring** with regard to a given RD, group of RD, or RD as a whole, by facilitating the **production of indicators**.



*thanks to ERNs and international experts' feedback and OD4RD current work (www.od4rd.eu)

ORPHACODES

ORPHACODES
Implementation in Health Records

with automatic transcoding to the generic terminology in use

To ensure that all RD are visible and that we leave no one behind

- to ensure adequate healthcare for ALL RD patients
  even when traveling abroad and in unplanned emergency situations
- to ensure adequate continuity of healthcare
- to improve evaluation and compensation of possible disabilities
- to facilitate health planning
- to contribute to assessing healthcare performance, resource allocation and outpatient and inpatient-related activities on rare diseases, including for ERN evaluations

To allow data to be interoperable between systems and countries

ORPHAcodes are aligned to the main generic terminologies so that you can retrieve the corresponding generic code (transcoding) without additional burden

- to facilitate data re-use in systematic collections (registries) for research and innovation
- to facilitate identification of patients suitable for integration in a clinical study including those undiagnosed
- to achieve critical mass of data
  and so contribute to economical and societal impact of the specific RD or group of disease or all RD and contribute to epidemiological knowledge and/or provide exact figures for pharmaceutical developments or negotiations as well as research.

Tools and services are available to facilitate technical implementation and alleviate burden on coders

www.od4rd.eu

ABOUT ORPHACODES

"Addition of ORPHAcodes to eHealth records would allow RD cases to be reported and costed easily."

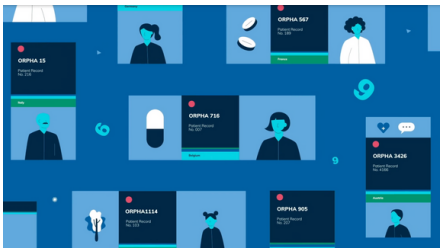
Gunne et al. 2020. A retrospective review of the contribution of rare diseases to paediatric mortality in Ireland

"ORPHAcodes were found to be a versatile resource for the coding of RD, able to assure easiness of use and inter-country comparability across population and hospital databases."

Mazzucato et al. 2023. ORPHAcodes use for the coding of rare diseases: comparison of the accuracy and cross country comparability

"ICD-10-ES codes have not enough specificity to identify rare diseases. Direct mapping between ICD and ORPHAcodes or the integration of ORPHAcodes at the healthcare system for diagnoses codification would enable better detection and epidemiological analysis of rare diseases"

Rico et al 2021. The interoperability between the Spanish version of the ICD and ORPHAcodes: towards better identification of RD



" Cross referencing between ICD-10 and ORPHAcodes may be adopted in different healthcare datasets for international comparison."

Chiu et al 2018. Healthcare burden of RD in Hong Kong adopting ORPHAcodes in ICD-10 based Healthcare administrative datasets

ORPHAcodes are the recommended code to trace RD diagnoses
Guidelines on Patient Summary, Release 3.2, Mar 2022 & Recommendation on ways to improve codification for rare diseases in health information systems (2014) European Commission Expert Group on Rare Diseases 2014 & European Common Semantic Strategy 2019

ORPHAcodes are listed in the set of common data elements for Rare Diseases Registration
released by the EU RD platform to ensure interoperability between registries

ORPHAcodes are identified as a best practice
by the Steering Group on Health Promotion, Disease Prevention and Management of Non-Communicable Diseases

X-eHealth project work included ORPHAcodes in the MDS required for the ePS specifications (D5.6-Refine-PS-functional-specifications-to-account-for-eHN-Guidelines-and-rare-diseases)