ORPHAcodes Implementation in The Netherlands

OD4RD2

Hospital Managers perspective

April 2023

Erasmus MC

Collaboration between Dutch Hospital Data & Dutch (medical university) hospitals

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- Member of EUHA network on rare diseases & ERNs

Need for better registration of rare diseases

Orphacodes since 2015 used in the acknowledgment of expert centers for rare diseases by Dutch Government. But…:

- Insufficient data available on rare diseases. Current registration not specific enough.
- One uniform registration on national level was missing.
- Need for better registration much expressed by professionals & expert centers, without creating extra administration.
    - E.g.: ‘how many patients on rare disease A have we treated?’
- Data necessary for research, registries, ERN collaboration, national collaboration, policies, exchange of data etc.

Using existing registration method in NL

- Dutch hospitals use nationwide standard list to register diagnoses in EHR (Diagnosethesaurus) with derivations to:
  - ICD-10
  - Codes for claiming healthcare costs
  - SNOMED CT
- Instruction to DHD in 2020: make derivation from Diagnosethesaurus to ORPHAcode. Start with ORPHAcodes from international ORPHA-SNOMED mapping tabel.
  - 1,850 existing diagnoses got derivation to an ORPHAcode
  - 3,100 new diagnoses added to Diagnosethesaurus including derivation to the ORPHAcode

- Doctors register their patients the way they are used to (with the Diagnosethesaurus), but ORPHAcodes can be derived from the patient records → no double/extra registration.

EXAMPLE

- Diagnosethesaurus 14631 '*Amyotrofe laterale sclerose*'
  - ICD-10: G12.2 '*Motor neuron disease*'
  - SNOMED: 86044005 '*Amyotrophic lateral sclerosis*'
  - Codes for claiming healthcare costs:
    - 0522 '*ALS*' for Neurology
  - ORPHA:803 '*Amyotrophic lateral sclerosis*'

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Nationwide database on rare diseases: ORPHA-viewer

- All Dutch hospitals supply data to DHD on the patients they treat (*LBZ).* DHD uses this data to give information back to the hospitals (benchmarks/dashboards), to governmental institutions, and others.
- Instruction to DHD in 2022 to develop a viewer on this database to give insight in number of patients with rare diseases: ORPHA-viewer (*in development)*
- Data supply from hospitals to DHD on rare diseases not yet 100% and validated
- ORPHA-viewer will be used in future by hospitals to supply data to the Ministry of Health, but also to exchange data within the ERN's.

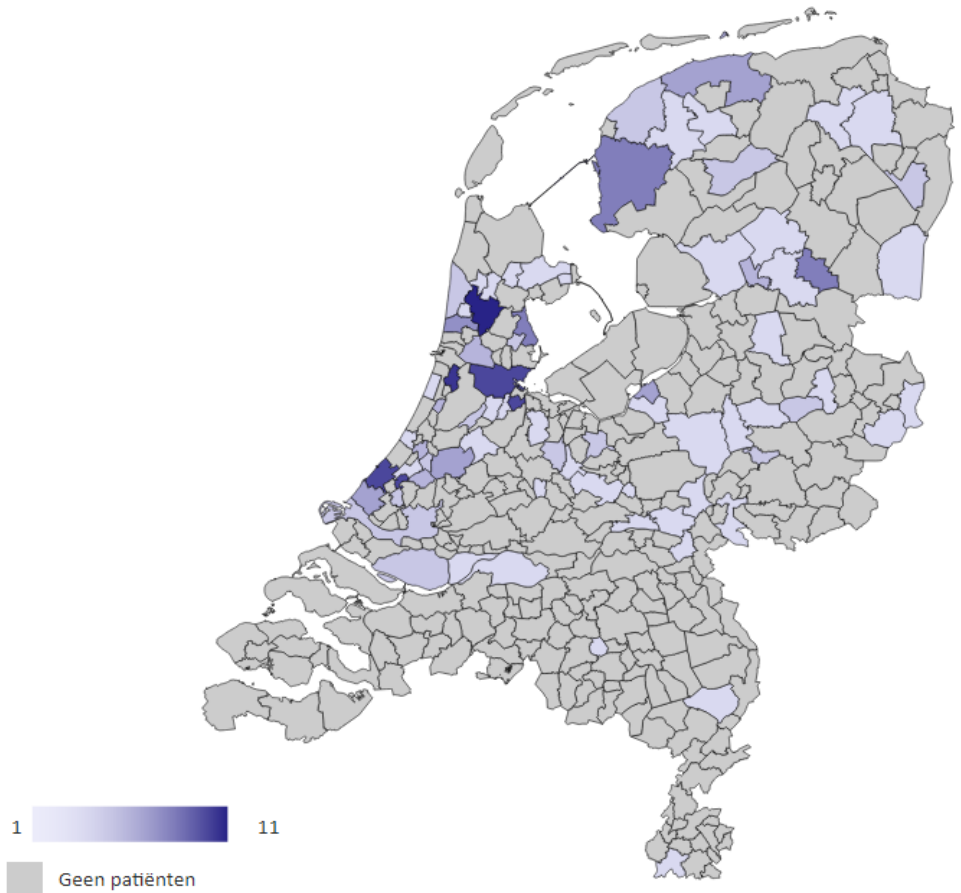
IN DEVELOPMENT

Aantal patiënten landelijk

168

Aantal patiënten per gemeente

Eigen zorginstelling Landelijk



RESET FILTERS

Periode

Multiple selections

Diagnose

Zeldzame systemische of reum...

60030

Zeldzame systemische of ...

60030 - Syndroom va...

60030 - Syndroom van Loeys-Dietz

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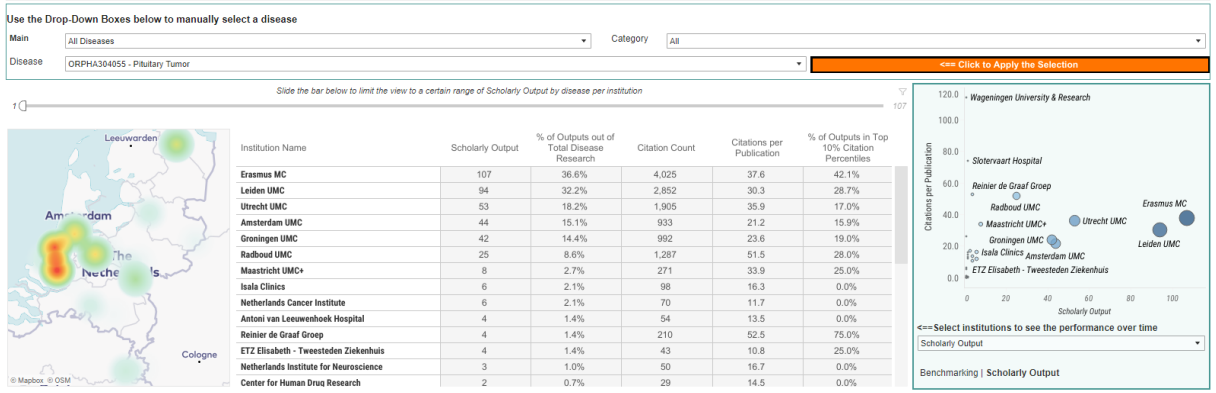
OrphaDiagnose	Man	Vrouw	Total
903 - Ziekte van Von Willebrand	671	1.385	2.056
Jaren 1920–1929	1	1	2
Jaren 1930–1939	3	21	24
Jaren 1940–1949	45	74	119
Jaren 1950–1959	53	137	190
Jaren 1960–1969	49	136	185
Jaren 1970–1979	42	142	184
Jaren 1980–1989	66	234	300
Jaren 1990–1999	52	245	297
Jaren 2000–2009	116	209	325
Jaren 2010–2019	199	152	351
Jaren 2020–2029	45	34	79
Total	671	1.385	2.056

Need to develop a method to identify research output on rare diseases

- Research output part of acknowledgement as an expert center
- Patient numbers don't tell us everything about who is de expert
- Get more grip on research portfolio and position within research field
- Finding research collaboration partners is of added value because of limited expertise on rare diseases
- Information valuable for professionals, hospital managers and patient (organizations)

Solution: analytics tool developed in collaboration between Elsevier and NFU (federation Dutch University Hospitals)

- ORPHAcodes used to match research output to rare diseases
- Result: Over 40.000 articles identified (period 2011-2022, output from Dutch institutions, Scopus database)



	Scholarly Output	% of Outputs out of Total Disease Research	Citation Count	Citations per Publication	% of Outputs in Top 10% Citation Percentiles
Spermatocytic Seminoma	9	100.0%	316	35.1	44.4%
Somatostatinoma	7	100.0%	67	9.6	0.0%
Muenke Syndrome	8	100.0%	133	16.6	12.5%
Pediatric Multiple Sclerosis	5	100.0%	680	136.0	40.0%
Megacystis-microcolon-intestinal Hypoperistalsis Syndr..	5	100.0%	135	27.0	60.0%
Jejunal Neuroendocrine Tumor	7	100.0%	67	9.6	0.0%
Allan-herndon-dudley Syndrome	28	96.6%	773	27.6	39.3%
Pure Or Complex X-linked Spastic Paraplegia	28	96.6%	773	27.6	39.3%
Peripheral Hypothyroidism	28	96.6%	773	27.6	39.3%
Trichothiodystrophy	14	93.3%	806	57.6	7.1%
Saethre-chotzen Syndrome	12	92.3%	251	20.9	8.3%
Omenn Syndrome	11	91.7%	307	27.9	45.5%
Postpartum Psychosis	19	90.5%	556	29.3	47.4%
Glycogen Storage Disease Due To Acid Maltase Deficien..	113	90.4%	2,581	22.8	25.7%
Congenital Alveolar Capillary Dysplasia	9	90.0%	307	34.1	44.4%
Lysosomal Glycogen Storage Disease	115	89.1%	2,608	22.7	25.2%

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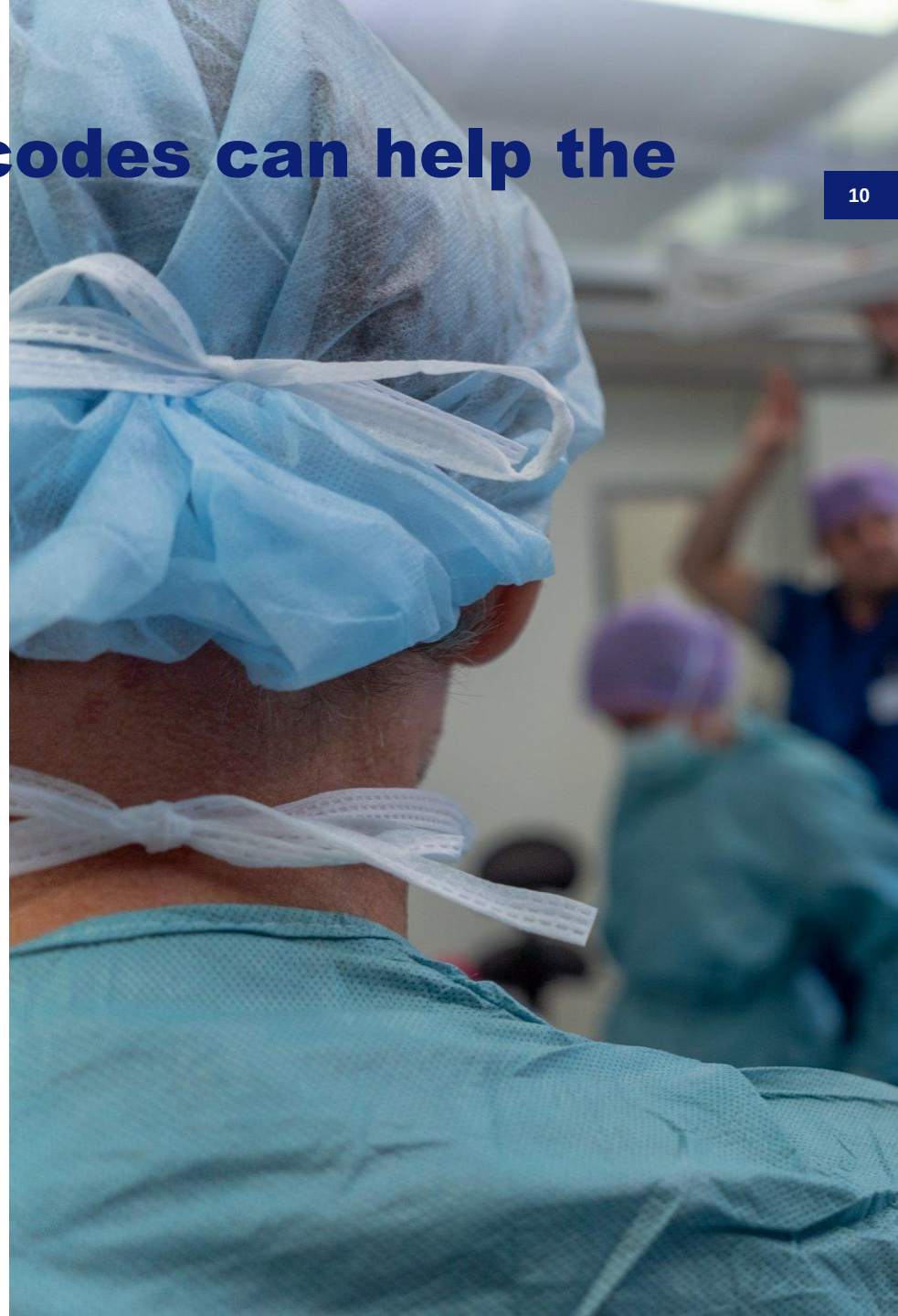
Implementation comes with some difficulties

- Complexity of ORPHAcodes structure (same disease in many orphathrees, on many positions): Difficulty of clustering ORPHAcodes (and therefore patient numbers, publications etc.) to higher (group) level
- Dutch situation: Medical registration in the first place used for declaration (DRD structure). Depending on scientific associations to include extra (billable) medical diagnoses.
- Working diagnosis not always updated in EHR by doctor
- Disease names in Orphanet not always the same as used within the medical field. Professionals search for a disease, not an ORPHAcode
- ORPHAcodes do not always (or already) fit with current registries; to specific or to general than the level needed for registry -> important to know where people can go to with remarks & requests

Further implementation of ORPHAcodes can help the rare disease field

- Number 1 : Create more & specific insight for professionals, hospital managers and patients (patient volume, research output, health care expenditures, origin of patients, capacity use, origin, referrers etc.)
- Possibility to connect it to other data on a rare disease level
- Making it easier to submit data for national acknowledgment and participation in ERNs
- Stimulate (research) collaboration by the possibility to exchange data and gain more insight
- Hopefully downsize administration burden (e.g. no manual lists)
- Etc.



Questions?