Orphanet Database analysis on RD coverage by ERNs

OD4RD
Orphanet Data For Rare Diseases

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OBJECTIVES

- Perform a quantitative analysis of ERNs coverage of RD in Orphanet classifications
- Update the 2019 analysis after the 2021 ERNs extension
- Set the ground for more detailed, qualitative analysis on:
  - Quantitiative and qualitative gaps in RD coverage
  - Quantitiative and qualitative overlaps / complementarities among ERNs
- Inform ERN coordination, BoMS and the EC's ERNs strategy
- Contribute to the JARDIN JA

METHODOLOGY

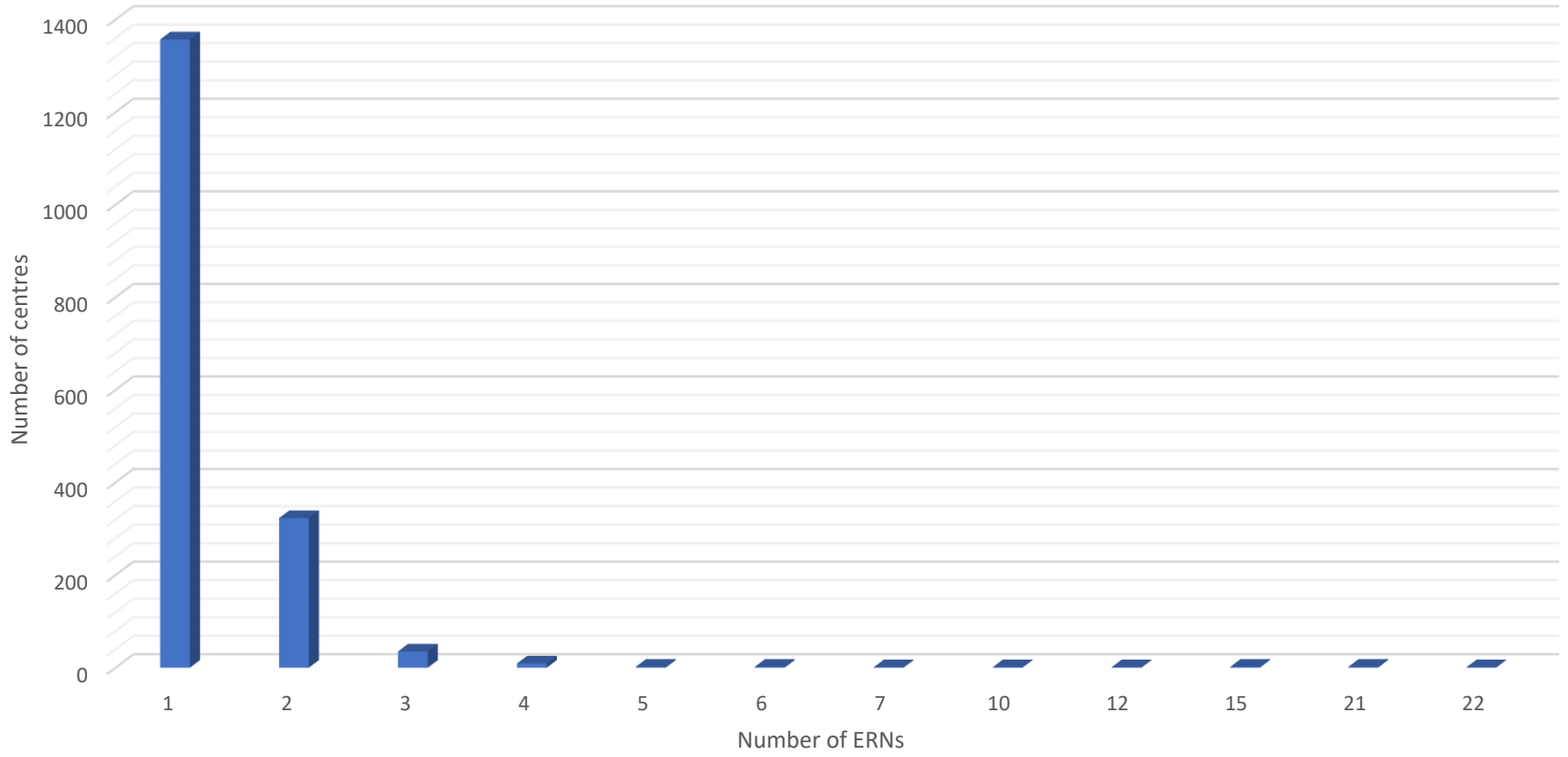
- Sources
  - Application files (EC): thematic groups, sub-tematic areas, exemplar RD
  - Orphanet classifications of RD
  - Orphanet ERNs centres database linked to the classifications (~99% complete)
- Steps
  - ERNs' centers data collection and mapping to the Orphanet classification of RD
  - Data extraction and preparation from the Orphanet database
    - Combining expert centres DB and Orphanet classifications files
    - Mapping thematic/subthematic areas to ORPHAcodes

WORKING HYPOTHESIS

- When mapping a thematic group or a sub-thematic area to an ORPHAcode representing a group of disorders, all the disorders within the group are considered as covered by the ERN, even though they are not mentioned in EC files.
- Justification:
  - If only mentioned RD were considered covered, most RD would have been in a « gap »
  - There is no real expert center on the rarest RD, but patients would benefit for coming to the ERN that is expert on the group the RD belongs to.

RESULTS

- 1,736 expert centres linked to ERNs
  - 78% linked to 1 ERN
  - 26 generic centres withdrawn for analysis purposes



AGGREGATED RESULTS

- For 12 ERNs, the coverage of RD in the related Orphanet classification groups was complete or almost complete
  - Complete: **GENTURIS , TransplantChild , ERNICA , Euro-NMD , eUROGEN, EURACAN**
  - Almost complete:
    - Few RD / clinical groups of RD not explicitly mentioned but could confidently be attributed to the ERN (pending validation)
    - **EpiCare , Rare Liver , RITA , ERN-EYE , EuroBloodNet , ITHACA**
- By ERN verifications are needed to confirm / infirm our assumptions
- Orphanet classification structure revisions identified as needed
  - i.e. ERN-Liver…

AGGREGATED RESULTS

- For 4 ERNs, the coverage of RD in the related Orphanet classification groups was incomplete but groups left out are covered by other ERNs
  - **ERKnet , CRANIO , ERN-LUNG , MetabERN**
- For 8 ERNs (needs confirmation/infirmation by ERNs):
  - RD groups left out are likely to be covered but not mentioned in the EC files
    - Needs confirmation by ERNs: **PaedCan, ERN-Skin, BOND**
  - RD groups left out and not explicitly covered by another ERN: true gaps? extensions possible within the current expert centers in the ERN?
    - **ENDO-ERN , Guard-Heart , ReCONNET , ERN-RND , VascERN**

PRESUMED GAPS

- Certain benign tumors: Inconsistently covered by domain-specific ERNs
- Certain systemic (autoimmune) diseases
- Certain acquired RD (while genetic counterparts are covered)
- Rare dyslipidemias
- Rare neurological infectious diseases
- Rare sleep disorders
- Rare pain disorders including Rare headache
- Rare autonomic nervous system disorder
- Medullar disease
- Prion diseases
- CNS Injuries
- Non-inflammatory vasculopathy

NEXT STEPS

- Confirm / correct this preliminary analysis
  - Per ERN discussions
  - Secondarily: identify RD groups for collaboration in revising the classifications
- Start the qualitative analysis
  - Investigate the already identified overlaps
    - Is it translating complementarity of approaches?
    - Is it due to the double/multi- ERN centres?
  - Share the results with JARDIN JA:
    - To understand the consequences in terms of patients pathways
    - To understand the consequences in terms of ERN monitoring