ERICA

European Rare Disease Research Coordination and Support Action

ERICA: WP3 Patient centred research and Patient Reported Outcome Measures

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European Commission

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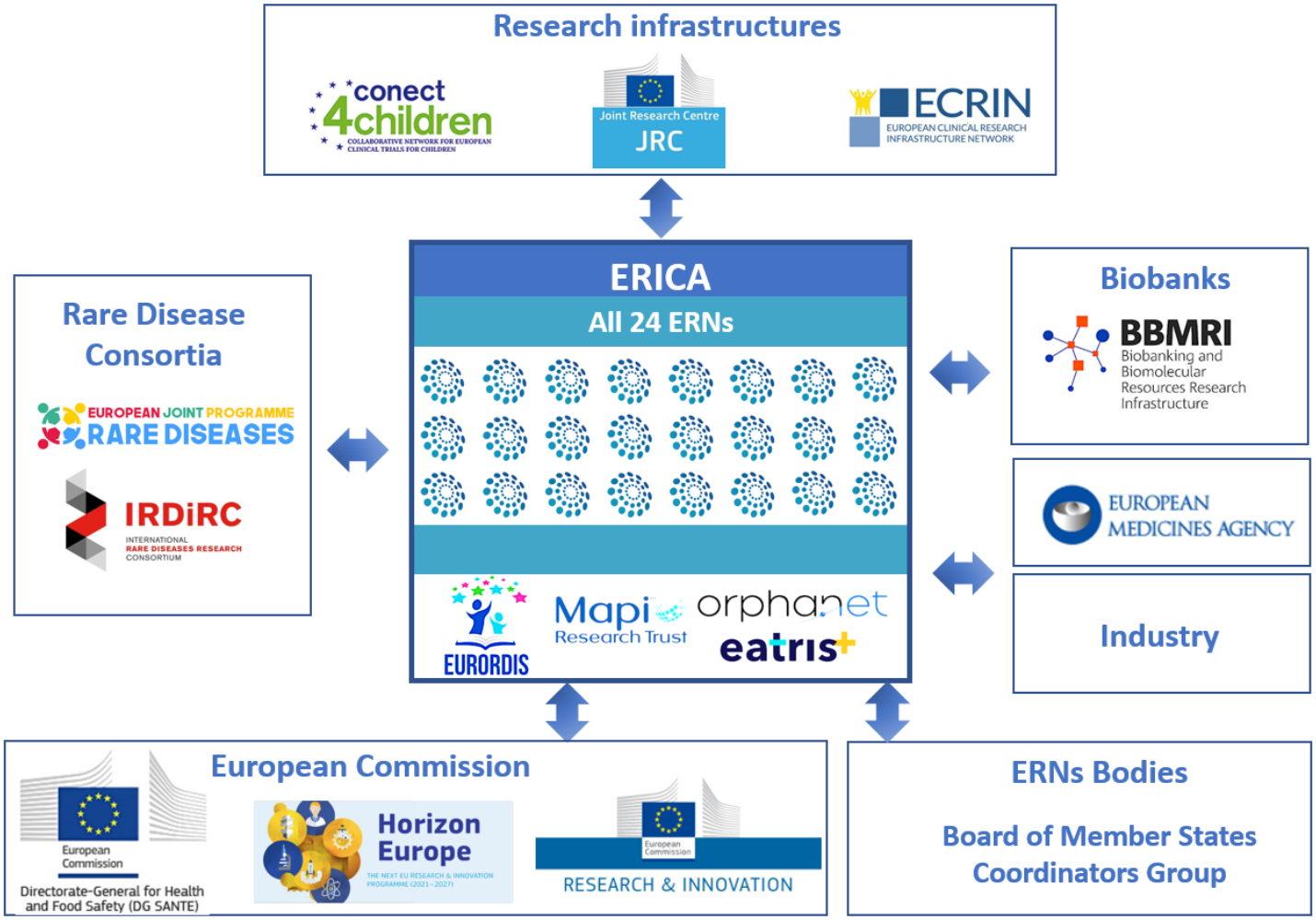
European Reference Networks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COLLABORATIVE NETWORK FOR EUROPEAN CLINICAL TRIALS FOR CHILDREN
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EUROPEAN JOINT PROGRAMME RARE DISEASES
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Types of Clinical Outcomes Assessments (COAs)

- **Patient-reported outcomes (PRO):** measurements based on data provided by patients, or proxies, regarding their health condition.
- **Clinician-reported outcomes (ClinRO)**: based on a trained health-care professional's report following observation of a patient's health condition.
- **Observer-reported outcomes (ObsRO):** measurements based on an observation by someone other than the patient or a health professional who is in a position to regularly observe and report on a specific aspect of the patient's health.
- **Performance outcomes (PerfO):** measurements based on a task performed by a patient according to instructions administered by a health care professional.
- **Biomarkers:** physiologic, pathologic or anatomic patient characteristics measured by an automated process or algorithm as an indicator of normal biologic processes, pathologic processes, or biological responses to a therapeutic intervention

IRDiRC Report on Patient-Centered Outcome Measures. Initiatives in the Field of Rare Diseases. February 2016

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WP3: Patient centered research

- to create a central repository of validated common and domain specific RD PCOMs/PROMs for ERNs
- to define priority areas for future PCOMs/PROMs development
- to support ERNs in the implementation of validated instruments for PCOMs/PROMs

PCOM: Patient Centered Outcome Measures
PROM: Patient Reported Outcome Measures

WP3: Patient centered research

- **Task 3.1: Strategic steering of identification, development and implementation of patient-centered outcome measures in ERNs-lead clinical research** (VHIR, INSERM/Orphanet);

- **Task 3.2: State of the art of available PCOMs/PROMs and overlap/gap analysis** (INSERM/Orphanet; Mapi Research Trust, VHIR, EURORDIS, ERNs coordination teams);

- **Task 3.3. Supporting ERNs on implementation of validated instruments for PCOMs/PROMs** ((VHIR, INSERM/Orphanet, SERMAS, interested ERNs).

Task 3.2. State of the art of available PCOMs/PROMs and overlap/gap analysis

- Orphanet and Mapi Research Trust will map existing validated tools for PCOMs/PROMs in RDs using ORPHANET alignments of RD nomenclature with other terminologies, including the expansion to functional consequences of RDs.

- Literature review and Survey among ERNs to identify succesful use cases of PCOMs implementation / on-going initiatives for PCOMs development or validation

Task 3.2. State of the art of available PCOMs/PROMs and overlap/gap analysis

- Categorize RD given their **functional consequences and impact on daily life** - based on ORPHANET Disability Questionnaire
- Questionnaire completed by RD experts: clinicians and patients representatives during phone interviews
- **Functional consequences** are indexed/annotated according to their **severity, frequency and temporality**

	Yes	No	**Permanent limitation (activity limitation)** **degree of severity** (**L:** low, **M:** moderate, **S:** severe, **C:** complete, **U:** unspecified) **frequency** (**VF:** very frequent: >80%, **F:** frequent: 30-80%, **O:** occasional: <30%)	**Delay in acquisition of a skill** **degree of severity** (**L:** low, **M:** moderate, **S:** severe, **U:** unspecified) **frequency** (**VF:** very frequent: >80%, **F:** frequent: 30-80%, **O:** occasional: <30%)	**Transient limitation (acute episodes, …)** **degree of severity** (**L:** low, **M:** moderate, **S:** severe, **C:** complete, **U:** unspecified) **frequency** (**VF:** very frequent: >80%, **F:** frequent: 30-80%, **O:** occasional: <30%))	**Loss of an ability limiting the activity** (e.g. degenerative disease)
examples:						
Are they impaired in their ability to receive a message?	☒	☐	*L (F), S (O)*		*C (F)*	☒
Are they impaired in their ability to learn?	☒	☐	*M (F)*	*L (O)*		☐
1 – Are the patients affected in their abilities to **understand and learn**?		☐				
if your answer is « Yes », please reply to the questions below						
if your answer is « No », please go directly to question 2						
Are they impaired in **seeing/watching**?	☐	☐				☐
Are they impaired in **hearing/listening**?	☐	☐				☐
Are they impaired in **learning**?	☐	☐				☐
Do they have difficulties **acquiring language** (oral language/ sign language)?	☐	☐				☐
Do they have difficulties **learning to read** (text/Braille)?	☐	☐				☐
Do they have difficulties **learning to write** (text/Braille)?	☐	☐				☐

Task 3.2. State of the art of available PCOMs/PROMs and overlap/gap analysis

- Starting point: systematic accumulation of standardized information based on the Orphanet disability questionnaire (ICF adopted)

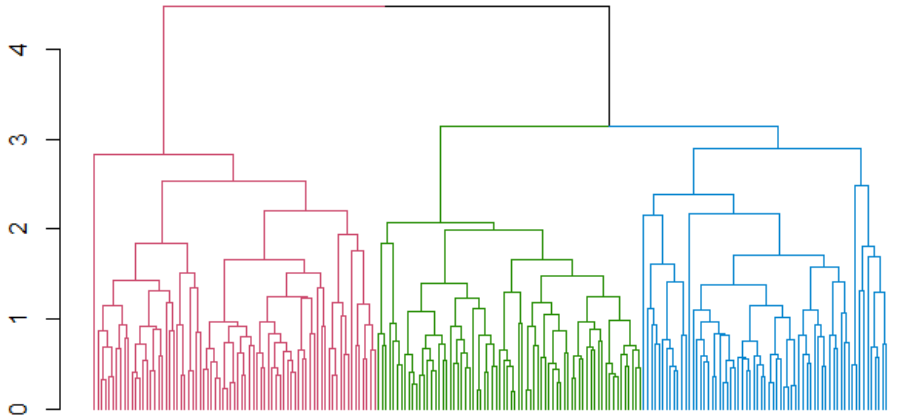
- 566 RDs coded for impact on function through a full set of 113 items grouped in 10 principal subjects describing patients' functional limitations:

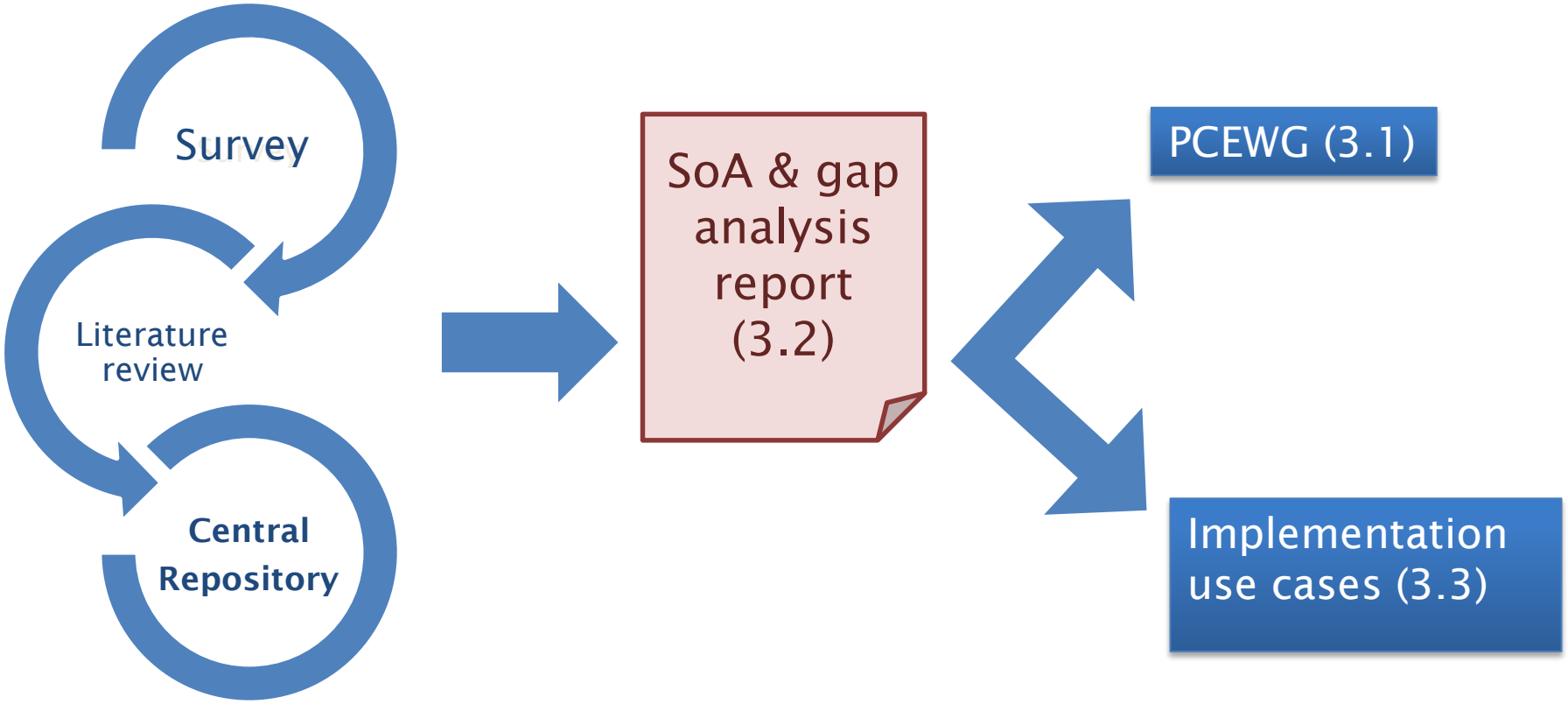
1. Understanding
2. Communication with others
3. Motor skills
4. Self-care
5. Sleeping/Staying awake
6. Temperament and behaviour
7. Moving around
8. Interpersonal skills
9. Daily activities
10. Social life

Task 3.2. State of the art of available PCOMs/PROMs and overlap/gap analysis

- Categorize rare disease given their functional consequences and impact on daily life- based on ORPHANET Disability Questionnaire
  - Clusters of diseases sharing similar phenomenology
  - Functions and group of functions relevant to Clusters
  - Describe Patterns of functional impairment Shared within clusters

- Define coding rules of COA instruments (still based on these functions)
- Prioritize targeted functions to be captured
- Identify instruments that match the needs and gaps





PCEWG: Patient Centered Expert Working Group

Thank you!