



## RECOMMENDATIONS FOR IMPROVING QUALITY OF RARE DISEASE REGISTRIES

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| Topics   | RECOMMENDATIONS   |
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| <b>Governance</b>  | <ul style="list-style-type: none"> <li>• define clear objectives;</li> <li>• engage with all relevant stakeholders, especially including patients representatives at early stage in the implementation of a registry;</li> <li>• establish a good registry team with clear role and responsibilities for different staff working members; is recommendable to include a person responsible for quality registry in the governance team from the start</li> <li>• ensuring compliance with (inter-) national and local regulation, develop rules and document for data ownership; data security measures, data access, informed consent it is recommendable to include a person responsible for quality registry in the governance team from the start.</li> </ul>   |
| <b>Data source</b>                                       | <p>The selection of primary data sources are critical for the success of the registry; they provide data of higher quality than secondary data sources; before incorporating a secondary data source into a registry, it is important to consider the legal and ethical feasibility of its incorporation and the potential impact of the data quality of the secondary data source on the overall data quality of the registry.</p> <p>Primary data sources are expensive and time consuming, you should consider the possibility of reuse of existing data from secondary sources.</p>   |
| <b>Data Elements, Case Report Form, Standardisations</b> | <p>The following step are recommended for defining DE:</p> <ul style="list-style-type: none"> <li>• determine what data is needed for the purpose(s) of the registry.</li> <li>• determine what information models and forms exist that can be reused.</li> <li>• determine what data comes from primary sources (requiring additional effort to collect) and from secondary sources (at the risk of lower data quality)</li> <li>• determine what data can be derived from other data, rather than being collected separately</li> <li>• determine whether data can be collected and stored in the clinical process (becoming data from secondary source)</li> <li>• determine whether data can be fed back to the clinical process</li> </ul> <p>Ensure and promote the use of standards in the registry system a) for diseases classification such as ORPHANET rare diseases Ontology (ORDO) and b) for phenotypes description (such as The Human Phenotype Ontology (HPO). Use of standards facilitates the data interoperability and availability.</p> |
| <b>IT Infrastructure</b>                                 | <p>Create free unprotected demos so non-registered users can explore the infrastructure. selecting a secure-by-design Information System infrastructure, technically accessible, including data storage, data management, data validation tools; establish Findable, Accessible, Interoperable and Reusable (FAIR) data including metadata.</p>   |
| <b>Data quality</b>                                      | <p>In order to address data quality, introduce quality assurance and quality control activities at different levels. Monitor regulatory data quality at central level and local and produce regular data quality report.</p>  |
| <b>Quality information</b>                               | <p>develop a defined statistical analysis plan describing the statistical techniques to be used in order to address the objective(s) of the registry; ensure data dissemination to different stakeholders: registry holders, patients, general public, decision makers and researchers.</p>   |
| <b>Documentation</b>                                     | <p>Developing and maintaining transparent and adequate documentation is essential for ensuring the quality and efficient operation of the registry. The detail of documentation may vary from registry to registry depending on the complexity of the registry.</p>   |
| <b>Traning</b>   | <p>ensuring proper and systematic training at all level, addressed to registry’s staff and data providers. Providing training in a systematic way and when changes occur.</p>   |
| <b>Data quality audit</b>                                | <p>Have an audit system including defined triggers initializing audit processes.</p>  |