

Título del Proyecto	Orphanet Data for Rare Diseases (OD4RD)
Nº de expediente asignado	GA nº: 101070531
Abstract	<p>Orphanet is a reference network for information and data on RD, representing 42 countries (including 25 MS), with its teams housed by institutions endorsed by national Authorities to conduct Orphanet related activities. In particular, Orphanet maintains a comprehensive RD nomenclature system aligned with several non-RD specific terminology resources, allowing for semantic interoperability in a context of heterogeneity of coding systems used in different countries. The nomenclature is also annotated with curated information on genes, epidemiological data, phenotypic traits, and functional consequences. This unique body of knowledge, completed by textual information, is delivered in both computable and human-readable formats. ERNs are end-users of the Orphanet nomenclature, but also active contributors to its development, since they concentrate clinical and scientific expertise as knowledge evolves. For this reason, a methodology was established to address ERNs' needs in terms of revision of the nomenclature during the OrphaNetwork Direct Grant (2018-2021), based on the collaboration with a number of ERNs. The Orphanet Data for Rare Disease project will build on Orphanet's specific expertise, and on its organisation as a long-lasting, well-established network, to fulfil the following general objectives: - To contribute to the generation of standardised, interoperable data on RD diagnosis for primary and secondary use, through maintenance of the Orphanet nomenclature of RD in collaboration with ERNs, and active support for its implementation in hospitals hosting ERNs, - To contribute to the harmonisation of data collection amongst various settings (health records, registries) and amongst countries, through dissemination of coding good practices at the source (health records, registries, etc). - To support evidence-based decision-making in the frame of the European staround ERNs, by providing an exploitable reference corpus of data and information on RD.</p>
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Enlaces:

<https://www.orpha.net/consor/cgi-bin/index.php>
<https://www.ciberisciii.es/areas-tematicas/grupo-de-investigacion?id=17811>
<https://www.ciberisciii.es/areas-tematicas/grupo-de-investigacion?id=17120>