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Orphanet-European Reference Networks collaboration

Paris, the 7th of June 2017.

Dear ERN coordinators,

It is with great pleasure that we formally invite you and your network to closely collaborate with Orphanet (www.orpha.net), the European database for rare diseases. By working together, we hope to coordinate our complementary activities so as to produce, improve and share information and data in the rare disease field.

As part of RD-ACTION (www.rd-action.eu), the European Joint Action on Rare Diseases, Orphanet is co-funded by the DG-Santé to:

- Produce the standard nomenclature of rare diseases (ORPHA nomenclature), with the aim to provide stakeholders with a common, controlled language to improve the interoperability between health information systems and databases and registries;
- Produce a database of scientific annotations for rare diseases, in order to provide the community with an up-to-date, added-value resource for re-use in healthcare and research;
- Produce comprehensive, up-to-date, expert-reviewed information on rare diseases, intended for all stakeholders, comprised of scientific data and texts;
- Publish a directory of activities related to RD in Member States, in particular the activities of European Reference Networks in the fields of care and research;
- Produce OrphaNews (www.orphanews.org), the RD-Action dissemination tool.

Orphanet and ERNs represent a common endeavour to improve rare diseases patients' lives in Europe, by increasing the knowledge and providing equal access to expertise.



It is therefore essential to synergise our work around the aforementioned missions. More precisely, we invite you to:

- Register the activities performed in the framework of your network (research projects, clinical trials, registries, diagnostic tests, centres of expertise) to ensure that they are visible and findable in the Orphanet website, and that your ERN is accurately represented, with all its constituent activities;
- Keep the community updated with your activities via OrphaNews, in the dedicated European Reference Networks section;
- Contribute to the improvement of the Orphanet nomenclature and encyclopaedia via our knowledge management web platform <https://curation.orphanet.org>. By registering as an expert, Orphanet can invite you to discuss and validate its scientific information in the future;
- Provide links on your ERN website to Orphanet (in particular the pages concerning the diseases covered by your network) in order for your users to find the most up-to-date information on the diseases you cover;
- Submit to Orphanet any clinical guidelines, review articles, articles for the general public, or other websites on the rare diseases you cover, that we could assess and consider for inclusion as a link in Orphanet.

You will find attached some practical documents containing all the necessary instructions and contacts, as well as the knowledge management web platform users' guide.

We are sure that this collaboration will be fruitful and will benefit the rare diseases community. Thank you very much in advance for your contribution.

A handwritten signature in black ink, appearing to read "AR".

Ana Rath

Director
Inserm US14 - Orphanet

Coordinator of RD-ACTION, European Joint Action on Rare Diseases 2015-2018
Coordinator of HIPBI-RD - Harmonizing information on phenomics for a better interoperability in the rare diseases field
Member of the Revision Steering Committee of the WHO's International Classification of Diseases (ICD11)
Scientific coordinator of Support-IRDIRC, Scientific secretariat of the International Rare Diseases Research Consortium