

[Inici](#) > RD-CONNECT: An integrated platform connecting registries, biobanks and clinical bioinformatics for rare disease research

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Description

Rare Diseases, Biobanks, Clinical Bioinformatics Despite examples of excellent practice, rare disease (RD) research is still mainly fragmented by data and disease types. Individual efforts have little interoperability and almost no systematic connection between detailed clinical and genetic information, biomaterial availability or research/trial datasets. By developing robust mechanisms and standards for linking and exploiting these data, RD-Connect will develop a critical mass for harmonisation and provide a strong impetus for a global trial-ready infrastructure ready to support the IRDiRC goals for diagnostics and therapies for RD in close collaboration with the successful A/B projects. It will build on and transform the current state-of-the-art across databases, registries, biobanks, bioinformatics, and ethical considerations to develop a quality-assured and comprehensive integrated hub/platform in which complete clinical profiles are combined with -omics data and sample availability for RD research. The integrated, user-friendly RD-Connect platform, built on efficient informatics concepts already implemented in international research infrastructures for large-scale data management, will provide access to federated databases/registries, biobank catalogues, harmonised -omics profiles, and cutting-edge bioinformatics tools for data analysis. All patient data types will be linked via the generation of a unique identifier (RD-ID) developed jointly with the US NIH. The RD-Connect platform will be one of the primary enablers of progress in IRDiRC-funded research and will facilitate gene discovery, diagnosis and therapy development. RD-Connect has the RD field at its heart and brings together partners with a strong track record in RD research (gene discovery and development of innovative treatments), as well as committed IRDiRC funding partners and representatives of all major international RD initiatives (EU/US/AU/JP) spanning patient organisations, research and public health, to maximise impact to RD patients

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