RD-Connect: an FP7 success story

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Summary

Although individually uncommon, rare diseases (RDs) collectively affect 6-8% of the population, around 30M people in the EU. RD research faces specific challenges. Since patients, clinical expertise and research communities are scarce and fragmented, data sharing between researchers is crucial.

To address this, in 2012 the European Commission awarded a €12M FP7 grant to RD-Connect to create an infrastructure bringing together multiple data types used in RD research into a common resource for researchers and patients.

In six years, RD-Connect developed an integrated platform bringing together analysis tools and different types of data needed in RD research into a common resource for clinicians and researchers worldwide.

The RD-Connect platform consists of three systems: Genome-Phenome Analysis Platform, Registry & Biobank Finder and Sample Catalogue, which are open to any RD.

RD-Connect overview

RD-Connect received from the European Commission an FP7 grant of €12M over six years (2012-2018) to create a global research infrastructure for rare disease research. RD-Connect is also a flagship project of the International Rare Disease Research Consortium (IRDiRC).

The RD-Connect platform consists of three systems:

- The Genome-Phenome Analysis Platform is not only a data repository, but also a fully functional genome-phenome analysis interface with a specific focus on diagnosis and gene discovery. It enables researchers and clinicians (even without bioinformatics training) to easily identify disease-causing genes and their matching cases across databases.
- The Registry & Biobank Finder is a global directory of RD-related patient registries and biobanks. It is unique in that it contains data for each registered patient/sample per each disease, including genetic data, which facilitates preparing clinical trials.
- The Sample Catalogue lets researchers browse sample collections and find detailed information about individual rare disease samples available in RD-Connect. The tool facilitates the reuse of scarce and valuable biosamples for research.

These three systems are open to any rare disease and accept submissions from users and databases around the world. In addition, RD-Connect has developed several bioinformatic tools for analysis and interpretation of omics data, e.g. VarIT, ONC-HC-Finder, Human Splicing Finder and more.

Through novel mechanisms for data linkage, remote resources that hold FondoXXible, Accessible, Interoperable and Reusable (FAIR) data can be linked in at a per-patient or per-sample level. Researchers can access pseudonymised datasets in RD-Connect, which are linked at an individual level.

The value of RD-Connect has been recognised by the European Commission’s Directorate-General for Research and Innovation and the International Rare Disease Research Consortium (IRDiRC), which has recognised it as a key contribution to the wider public sector RD research and has included RD-Connect in its impact report on improving the quality of life of RD patients.

Research achievements

RD-Connect has already contributed to advances in RD research and has published over 250 scientific publications and contributed to over 100 genome discoveries.

The Genome-Phenome Analysis Platform was used as the primary tool for analysis of 900 exomes that have been sequenced as part of the 17 research projects funded in the BBMRI-LPC (www.bbmmi-lpc.org) call in 2016.

The Genome-Phenome Analysis Platform is also used by Solve-RD (www.solve-rd.eu), a major new Horizon 2020 project to diagnose undiagnosed patients with 104 European rare diseases. RD-Connect has provided access to 19,000 exomes to RD-Connect. Thus, at that time, the platform was expected to hold around 30,000 exomes datasets.

The Registry & Biobank Finder has already found hundreds of RD patient registries, and allows users to browse resources covering over 300 different rare diseases. The Sample Catalogue enables researchers to find detailed information on individual samples available in RD biobanks. Since its launch in December 2017, it has already received data from 400M samples stored in the joint biobank of the EuroBioBank network and will soon include samples from other European biobanks.

These examples show how the reuse of infrastructure developed in one project to allow its inclusion in new initiatives maximises the value of European funding.

Collaborations

During its development, RD-Connect closely collaborated with two parallel EU-funded research projects NeuOnC and EUrOnC, which applied omics technologies to study rare neuromuscular and neurodegenerative and rare pediatri MR disorders.

Thanks to this fruitful collaboration, RD-Connect received admissions of omics data, while the feedback from end-users allowed RD-Connect to develop tools to suit the needs of RD researchers. RD-Connect supported both projects in gene discovery and data linkage, which facilitated diagnostic pipelines for both projects.

Similarly, the collaboration with EuroBioBank, the European network of biobanks which became the Flagship biobank of RD-Connect, was fundamental for the development of theSample Catalogue.

RD-Connect, along with NeuOnC and EUrOnC, are embedded in European collaborative projects such as IRDiRC and research infrastructures such as BBMRI and ELIXIR.

RD-Connect has also initiated collaborations with the European Reference Networks (ERNs), which connect RD experts and healthcare providers across the EU. RD-Connect is supporting the ERNs in their research goals and in creating FAIR patient registries. This collaboration will greatly increase the project’s impact on improving the quality of life of RD patients.

International recognition

The value of RD-Connect has been recognised by the international RD community. The European Commission’s Directorate-General for Research has included RD-Connect among its Research and Innovation success stories and promted it using in dissemination channels, publications and international events.

To further promote the role of RD-Connect in RD research, its impact has been assessed by EMBL-EBI, which has awarded the label ‘IRDiRC recognized resource’ to three RD-Connect outputs:

The FAIR Guiding Principles for sharing biocompens data and social challenges; how do we maximise the benefits and opportunities for rare disease research and how do we ensure that innovation and patient engagement are core to the process? The FAIR Guiding Principles for sharing datasets by 2019

Recommended