RD-CONNECT: a unified platform for rare disease research

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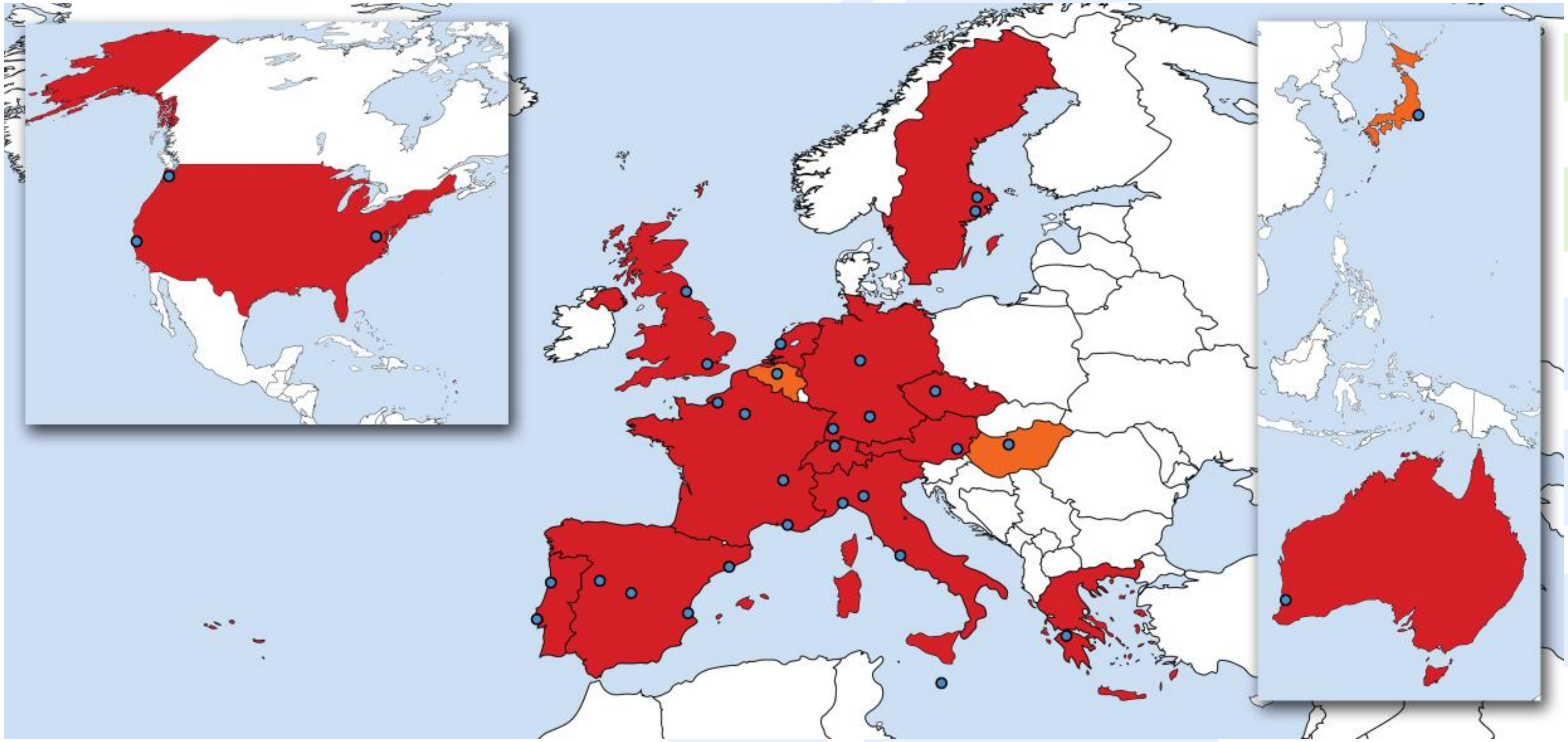
RD-Connect

- An integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research
- 12m euro EC contribution, 6 years
- Co-ordinated by Institute for Genetic Medicine, Newcastle University, UK
- Supports IRDiRC goals of Diagnosis for all rare diseases and 200 new therapies for RD – by 2020

Policy
Ethics
And
Life
Sciences

RD-Connect partners



27 full partners, 17 associated partners, 17 countries

RD Connect

RD-Connect central platform

- RD-Connect's central platform will make the data generated by IRDiRC projects rapidly available to the wider rare disease research community to accelerate the discovery of new genes, enhance the accuracy of diagnoses and facilitate the development of treatments
- Raw genomic data from collaborating projects will be securely deposited in the European Genome-Phenome Archive (EGA) before being processed through a standard pipeline to ensure cross-compatibility of data from multiple projects
- The processed data will be held in the central RD-Connect database, where it will be combined with other –omics data types plus phenotypic and biomaterial information
- Researchers approved by a data access committee will access data through a data coordination centre that enables comparison of datasets across projects and analysis with sophisticated bioinformatics tools

-omics

"Ever since the rise of genomics, the suffix '-omics' has been added to the names of many fields to denote studies undertaken on a large or genome-wide scale."

nature.com

Denotes study of particular subject in relation to genome, proteomics (proteins), nutrigenomics (food), pharmacogenomics (drugs)

Patient Participation

- Explore with patients and patient organisations their hopes, expectations and concerns regarding –omics research
- Scope the experiences and understanding of patients who contribute to registries and biobanks in rare disease
- Use findings to help scientists incorporate an understanding of patients’ hopes and expectations for the technologies into their work within RD Connect
- Manage Patient and Ethics Council (RD-PEC), which will answer questions on social, ethical and legal aspects of RD research from anyone with an interest

Why?

- Focus for dynamic dialogue between researchers and patients
- Inter-understanding increases uptake and acceptance of technologies
- Researchers know developments are relevant and needed
- Promotes high professional standards and ethical integrity
- Builds trust and confidence around new developments and technologies

First issues from patients

- Links between patients, families and researcher to include laboratory staff
- Education about projects and purpose – capacity build
- Challenge of explaining complexity – not just to lay people but researcher to researcher
- Incidental findings – hierarchy of consent
- Management of expectations, hope versus hype
- Fair representation, governance, opportunity to attend meetings
- Tools for reporting common issues/problems to wider audience
- Challenge ourselves to break out of usual (run of the mill) dissemination

Contacts

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