



## 21<sup>st</sup> Workshop

### EURORDIS Round Table of Companies (ERTC)

#### *"Sharing Rare Disease Patient Data: Translating Principles into Action"*

**Tuesday 30 September, 2014 (9:00 to 16:00)**

**Barcelona, Spain**

#### **CONCEPT PAPER**

Over the past decade, workshops of the EURORDIS Round Table of Companies (ERTC) have facilitated discussions on the value of collaboration all along the treatment lifecycle including clinical trials, regulatory requirements and health technology assessment. Today, it is clear that partnership is required not only downstream but also upstream the orphan medicinal product development process. In 2013, the 20<sup>th</sup> ERTC Workshop paved the way in underscoring the importance of future synergies between the research and medicines development agendas to ultimately support development-related investment decisions and well-designed clinical trials.

**Upstream**, the ability to link clinical and genetic data is crucial to translational research. This link contributes significantly to the identification of genes associated with diseases, the understanding of the frequency of genetic variants in populations and a better understanding of the reasons for drug reactions (both positive and negative). Currently, however, the integration of these two data types is limited by the fact that key infrastructures housing them (patient registries, biobanks, and genetic research databases) are not interoperable and thus not able to be connected. First steps to overcome such hurdles on the international level began with the establishment by the European Commission Health Research Directorate and the US National Institutes of Health of the International Rare Disease Research Consortium (IRDiRC) requiring that data and research results funded under this umbrella are shared among IRDiRC research projects and made publicly available to the broader community. Two initiatives already contributing toward this international objective include:

1) The Directorate General for Health and Consumers (DG SANCO) and the Joint Research Centre (JRC) agreement to join competences and develop a European Platform on Rare Diseases Registration. This platform will provide a central access point for all registry data on rare diseases, to act as a 'hub' improving access to patient registries, as well as to promote interoperability between



registries. A further important goal will be to improve data comparability, reliability and harmonisation among rare diseases registries throughout Europe. EURORDIS has and continues to work closely alongside this joint initiative to help implement proposed scenarios.

2) The RD Connect project, an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research. By facilitating the mechanisms and standards for linking and exploiting existing data and new data generated in related rare disease research projects, RD-Connect will develop a critical mass for harmonisation and provide a strong impetus for a global “trial-ready” infrastructure that is set to support the IRDiRC goals for complete diagnostics and 200 new therapies for rare diseases by 2020. Because of its direct involvement in a number of projects supporting the development of RD research infrastructures, EURORDIS is currently contributing to most aspects of the work in RD Connect on the operational and governance levels.

Numerous additional partners have joined IRDiRC. Patient representatives in this group have regularly demonstrated their role in bridging the translational gap by driving data sharing and integration through a patient-centred approach and assuring optimal efficiency and transparency.

To go beyond simply setting up new technologies and truly achieve impact and translating basic and clinical research findings into improved care and therapeutic solutions for the benefit of patients, close collaboration with the medical device and orphan drug developers is also essential **downstream**. Listening to the needs and concerns of the private sector around clinical trial preparation, development of drugs for the market and commercial aspects of biobanks, registries, diagnostic tests and standards will help drive the implementation of policies that take into due consideration all stakeholders’ needs. Furthermore, data collection and patient registration will be a key component of any European Reference Network to be established in the coming years and therefore, the rare disease community at large needs to reflect on how to cooperate and integrate these aspects, including data sharing, into the overall networking system.

Regulatory bodies and European legislation will strongly affect such collaborations both in the non-competitive and competitive space. Three fundamental Regulations (the General Data Protection Regulation and the Clinical Trials Regulation, as well as the In Vitro Diagnostic Medical Devices Regulation) concerning the 2020 IRDiRC goals are currently under discussion or just finalised in the EU legislative process. At the same time, the Cross-Border Healthcare Directive has now been transposed into national legislation and gives a legal basis to both the creation of the European Reference Networks and the improved European collaboration on Health Technology Assessment.



The many points of intersection between these different pieces of legislation must be better elaborated from a practical point of view, while maintaining a strong focus on the ethical considerations important to society.

Through the Patient Registry Survey conducted by EURORDIS, within the EPIRARE Project, on patients' experience and expectations in the field of patient registries, rare diseases patients have expressed an overwhelming support for 1. The creation of a common European infrastructure supporting registration and data collection and 2. The establishment of registries aimed at supporting healthcare and social planning, as well as generating knowledge and information. In order to increase patients' trust towards registries, they expect transparency in both using registry data and sharing the results of research projects.

A general consensus amongst rare diseases patients on the level of protection required for their personal data when balanced with the benefits of encouraging translational research, is particularly difficult to reach: on the one hand, sharing of scarce data is an absolute priority in order to foster research and facilitate translational projects in the rare diseases field, while on the other hand, patients living with rare diseases are in a vulnerable position as they are much easier to identify (e.g. through cross-cutting of databases) than patients with common conditions. The balance between protecting personal data and facilitating research into rare diseases is a particularly difficult one to strike.

Different scenarios are possible to realistically achieve improved diagnostic capabilities for all rare diseases patients and significantly increase treatments options. Improvement in rare disease therapies is tightly linked to a continuum of data-generation, a process which starts at pre-clinical stages and continues throughout post-approval phases. This long term process helps understanding the natural history of the disease, defining standards of care, identifying meaningful, measurable and validated endpoints, designing clinical trials protocols, etc.

In this context, collaboration amongst all the stakeholders represented at this 21<sup>st</sup> ERTC Workshop is a key condition for success. The technical, legal and ethical nature of acceptable models for data sharing and integration, the issues to consider when establishing such models and the good practices required to enhance the success of patient-centric efforts will be the focus of this workshop.