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Original Article

Biobanking: towards increased access of biomaterials in cystic fibrosis. Report on the pre-conference meeting to the 13th ECFS Basic Science Conference, Pisa, 30 March-2 April, 2016

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1. Introduction

Cystic fibrosis (CF) is caused by mutations in the *cystic fibrosis transmembrane conductance regulator* (*CFTR*) gene. With an incidence ranging from 1:2500 to 1:6000 across European countries, it is the most frequent life-shortening hereditary rare disease (RD) in Europe. Over the last decades, tremendous progress has been achieved in understanding CF disease mechanisms as well as how CFTR loss-of-function can be restored through pharmacological and genetic interventions [1]. Apart from the mutations in the *CFTR* gene, non-CFTR genetic modifiers and environmental factors are increasingly being recognized to impact CF disease expression. Nevertheless, how these factors determine individual CFTR residual function and disease expression remains mostly unknown [2]. Remarkable clinical improvements can be achieved upon restoration of CFTR function by CFTR modulators, but considerable heterogeneity in response to CFTR modulators is observed [3–7]. Similar to the heterogeneity in disease phenotype, the inter-patient difference in efficacy of treatment is likely influenced by many genes, whose protein products mediate complex interactions with CFTR (i.e. modifier genes) [8,9] as well environmental factors [10].

Despite this progress, treatments that can effectively restore CFTR function to levels of healthy controls remain lacking. More effective CFTR modulators and a better understanding of interactions between CFTR, other disease modifiers and CFTR modulating drugs for each individual are needed if we aim to develop effective treatments for all subjects with CF.

Biobanks are a valuable research resource to speed up drug development as well as to develop evidence-based patient care. Different definitions of biobanks exist, but the one used at this preconference meeting was ‘an organized collection of human biological materials and associated information stored for one or more research purposes’ that was adapted from Fransson et al. [11].

The extent to which biological materials and information are included in biobanks and how these are used vary among distinct

Abbreviations: BALF, broncho-alveolar lavage fluid; BBMRI-ERIC, Biobanking and Biomolecular Resources Research Infrastructure — European Research Infrastructure Consortium; BSWG, Basic Science Working Group; CF, cystic fibrosis; CFF, Cystic Fibrosis Foundation; CFTR, cystic fibrosis transmembrane conductance regulator; CS-ELSI, common service—ethical, legal, social implications; ECFS-CTN, European Cystic Fibrosis Society Clinical Trial Network; ELSI, Ethical, legal, social implications; iPS (cells), induced pluripotent stem cells; LoI, letter of intent; MTA, material transfer agreement; RD, rare diseases; SOP, standardized operating procedure; TDN-CC, Therapeutic Development Network Coordinating Center; TNGB, Telethon Network of Genetic Biobanks.

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biobanks and significantly impact their overall usefulness for advancing research. Biological materials can range from dead-tissue biopsies to frozen collections of living primary cells. Information about these materials can be as limited as a few anonymous identifiers (e.g. CF pulmonary cells) to as detailed as linkage to individual clinical data in a patient registry. Dependent upon the informed consent, materials and data can be used for defined or yet undefined research purposes by academic or both academic and commercial parties.

2. The preconference meeting: 'biobanking: towards increased access of biomaterials in CF'

The meeting organized by the federation of CF Patient Organizations Cystic Fibrosis Europe (CFE) and the European Cystic Fibrosis Society (ECFS) focused on how biobanks for CF can be established so that their impact on patient care and their usefulness for research can be maximized. For the generation of biobanks for rare diseases (RDs) such as CF, an international approach is needed to ensure that sufficient disease heterogeneity is captured. Indeed, more than 2000 different *CFTR* mutations have been reported so far (Cystic Fibrosis Mutation Database www.genet.sickkids.on.ca/) and the distribution of these mutations throughout different populations across the globe is strongly influenced by ethnicity and geographical location [12,13]. Common challenges for European biobanks include: 1) providing quality standards and standardized operating procedures (SOPs) for material collection and storage; 2) ensuring that collected materials are effectively distributed; 3) defining what sort of governance is most useful and effective in the context of Europe and how long-term sustainability can be ensured; 4) accommodating cultural and legal differences in ethical, legal and social implications (ELSI) of biobanking across the various European countries; and 5) involving the public, patients and/or patient representatives in all aspects of the biobank project.

To launch the basis for an European CF biobank, members of national CF patient/parent organizations from The Netherlands, France, Belgium, the United Kingdom, Italy and Germany set up a pre-conference program to learn about established European biobank infrastructures for other RDs, ELSI aspects of pan-European transfer of biomaterials, development and use of CF biorepositories by the US CF Foundation (CFF) and an early inventory of existing CF biobanks in Europe. The presentations were followed by a forum discussion focusing on how to move forward towards building a European CF biobank infrastructure that optimally facilitates CF research and patient care.

3. Critical factors in establishing a network of biobanks in rare genetic diseases (by Dr. Chiuhui Mary Wang, Program Manager for Telethon Network of Genetic Biobanks, EuroBioBank and RD-Connect Biobanks work package)

The need to create networks of biobanks is of particular importance for rare and genetic diseases, because of the rarity of biological materials to drive research on understanding disease pathogenesis, validation of diagnoses and development of treatments. Fondazione Telethon, an Italian non-profit organization that supports research on genetic diseases, has been funding single disease biobanks since 1993. A need to create a biobank network was observed, and subsequently the Telethon Network of Genetic Biobanks (TNGB) was created in 2007 [14]. The main aims of the TNGB are to: 1) virtually centralize access to very rare samples and data; 2) minimize biases arising from heterogeneity in the sample quality by developing and sharing SOPs and policies; and 3) promote the role of biobanks with patient associations and foster their participation in developing procedures concerning data privacy, informed consent and sample usage [15]. Crucially, the aims were enabled by a shared IT infrastructure (http://biobanknetwork.telethon.it/). The workflow of biomaterials was also standardized to facilitate biobank management, including templates for informed consent and material transfer agreements (MTAs). Currently there are 11 biobanks in the network, and researchers have quick access to sample collections via the TNGB portal. The TNGB is governed by the directors of the biobanks and is supported by an advisory board consisting of biobanking experts, ELSI experts and patients' representatives. This Italian network also undergoes peer-review by the Telethon Biobank Committee with international biobanking experts.

In the European scenario, RD-Connect, an infrastructure project funded by the European Union's Seventh Framework Program since 2012, represents a major networking opportunity for RD biobanks (http://rd-connect.eu/platform/biobanks). This 6-year project aims to develop an integrated platform connecting databases, registries, biobanks, and clinical bioinformatics for RD research. Experience from TNGB was brought into RD-Connect, where one of the core objectives is to make biological samples and associated data from RD patients accessible and available to the scientific community by creating an online searchable sample catalog [16]. Networking challenges including standardization of datasets, biobank quality standards, and support material for new biobanks are also addressed [17]. Perhaps even more importantly, the platform is being developed with guidance from patient voice and ethical experts on ELSI related to data and sample sharing in RD [18,19]. EuroBioBank (http://www.eurobiobank.org/), the oldest network of RD biobanks founded in 2001, is currently being integrated as a part of RD-Connect [20]. The project also seeks collaboration between international partners such as the National Institute of Health and BBMRI-ERIC to ensure platform interoperability and sustainability.

In conclusion, there are IT tools and support allowing RD biobanks to connect. The critical factors remain the efforts of the biobanks to adopt standards and common policies, and their drive to operate in a high quality infrastructure to serve the scientific community.

4. ELSI aspects of pan-European transfer of materials and of access to data/materials by academic and commercial stakeholders (by Dr. Sara Casati, Common Service ELSI BBMRI-ERIC, Italy)

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