The Role of Biobanking in Rare Diseases: European Consensus Expert Group Report

Hanns Lochmüller,1 Ségolène Aymé,2 Francesca Pampinella,3 Béla Melegh,4 Klaus A. Kuhn,5 Stylianos E. Antonarakis,6 and Thomas Meitinger7

Biobanking is of high importance for research in rare diseases. There are >6,000 rare diseases with at least 30 million people affected in the European Union (EU). The European Commission (EC) has prioritized rare diseases in recent health and research programs. The rarity and diversity of rare diseases and their associated biomaterials harbor specific challenges and opportunities for biobanking requiring transnational collaboration and harmonization. Small collections or even individual samples may be extremely precious for research. Importantly, most rare disease biobanks work through the active participation of patients and patient organizations, and share benefits with them. This article gives recommendations related to rare disease biobanking reflecting consensus of an expert working group of the Biobank and Biomolecular Research Infrastructure program at a meeting in Munich on December 17–18, 2008.

T/his article reflects the consensus of an expert working group of the Biobank and Biomolecular Research Infrastructure (BBMRI) program who met in Munich, Germany on December 17–18, 2008. All authors participated in this working group. The last author (T.M.) convened and chaired the meeting, the first 2 authors (H.L. and S.A.) organized the manuscript.

Recommendations of the Rare Disease Working Group

Rare diseases are defined as affecting <1 citizen in 2,000. There are >6,000 rare diseases with at least 30 million people affected in the 27 European Union (EU) member states. The rarity and diversity of rare diseases pose specific challenges that need to be addressed at European Commission (EC) and Member States (MS) levels. The EC has prioritized rare diseases in health and research programs (see EC Communication “Rare Diseases: Europe’s challenge” on November 11, 2008). Rare disease biobanks have been recognized as important tools for research into the cause and treatment of the conditions, with biomaterials (including tissues and cell lines) playing a key role in translational research.

Rare disease biobanks share similar objectives and face similar challenges as other biobanks such as population biobanks. Rare disease biobanks would benefit from a sustained infrastructure as proposed by BBMRI. The rarity and diversity of rare diseases and their associated biomaterials result in a pressing need for transnational collaboration, high demand for expert input for quality control of biomaterial, and high need for the training and education of scientists using the biomaterials. Small collections or even individual samples may be extremely precious for research, and some may have direct relevance for patients’ health. Importantly, most rare disease biobanks work through the active participation of patients and patient organizations (sample collection, funding, operative level), and share benefits with them.

More than 100 rare disease biobanks exist in Europe and many of these have expressed their interest in joining BBMRI. They are listed in the Orphanet database.

In France, the introduction of quality standards led to a reduction in the total number of biomaterial collections.

1Institute of Human Genetics, Newcastle University, Newcastle upon Tyne, United Kingdom.
2INSERM, SC11, Paris, France.
3Scientific Office, Telethon, Milan, Italy.
4Department of Medical Genetics and Child Development, University of Pecs, Pecs, Hungary.
5Institute for Medical Statistics and Epidemiology, Technische Universität München, Munich, Germany.
6Department of Genetic Medicine and Development, University of Geneva Medical School Geneva, Switzerland.
7Institute of Human Genetics, Helmholtz Center, Munich, Germany.
Telethon (Italy) has a national plan in place for rare disease biobanking that assures high-quality standards through regular assessments. The Telethon Network of Genetic Biobanks connects 7 rare disease biobanks in Italy sharing a common IT infrastructure and acting as a single entity regarding the management of samples and data, SOPs, ELSI issues, and quality assurance. There are numerous, established research collaborations in rare disease networks on both a national level and a European level.

EuroBioBank is a European network of rare disease biobanks with a focus on neuromuscular disorders. EuroBioBank was financed by the EC (FP5) between 2003 and 2006; it has been sustained since and was cited by the IPTS/ESTO work group as a European model of coordination and of integration of Biological Resources Centres for the optimization and improvement of the use of human biomaterial at a European level.

The interoperability between biobanks and between biobanks and other information systems is not satisfactory as, until recently, there was no nomenclature of rare diseases available. Orphanet is now providing a comprehensive coding system in 5 languages, which is at the disposal of the International community and is cross-indexed with ICD10 and MIM codes.

**Summary**

1. Biobanks are highly relevant for rare disorders. Rare disease biobanks have many commonalities with other biobanks, but face additional challenges due to the rarity and the diversity of the conditions and biomaterials.

2. It is rather the quality of the biomaterials and of the associated information than their quantity (number of samples) that is critical in rare disease biobanking.

3. This necessitates the adoption by biobanks of an appropriate disease coding system that accommodates all rare disorders, such as the nomenclature developed by Orphanet.

4. Active participation of and benefit sharing with patients and patients’ organizations is pivotal in rare disease biobanking. The Telethon biobanks (Italy) may serve as a role model (prototype) for networking rare disease biobanks on a national level, while EuroBioBank may be a role model (prototype) for networking rare disease biobanks on a European level.

5. Rare disease biobanks have common objectives with BBMRI and recognize BBMRI as an important mechanism to secure the long-term sustainability of biobanks in Europe.

6. Rare disease biobanks may provide their experience and advanced solutions to BBMRI, some of which may be applicable to other biobanks.

**Address correspondence to:**

Dr. Hanns Lochmüller  
Institute of Human Genetics  
Newcastle University  
International Centre for Life  
Central Parkway  
Newcastle Upon Tyne NE1 3BZ  
United Kingdom  

E-mail: hanns.lochmuller@newcastle.ac.uk

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