BBMRI – WP3 meeting/rare diseases working group

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Introduction: The rare-disease working group met for the first time by invitation from T. Meitinger on December 17/18 in Munich and agreed on the following recommendations for BBMRI.

Recommendations of the rare disease working group for BBMRI

Rare diseases are defined as affecting less than one citizen in 2,000. There are more than 6,000 rare diseases with at least 30 million people affected in the 27 EU member states. The rarity and diversity of rare diseases pose specific challenges which need to be addressed at EC and MS level. The EC has prioritized rare diseases in health and research programs (see EC Communication "Rare Diseases: Europe's challenge" 11 November 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. 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 seven 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 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, 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 optimisation 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 five languages which is at the disposal of the International community, which is cross-indexed with ICD10 and MIM codes.

<u>In summary:</u>

- 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.
- 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.

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

▪ 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.

▪ 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.

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