Instituto_Nacional de Saúde Doutor Ricardo Jorge

September 2011

Abstracts

International Workshop

Biobanking for Health Research

_National Health Institute Doutor Ricardo Jorge, INSARJ
Lisbon, Portugal

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September 28-30, 2011
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Organizing Committee

Marta Barreto da Silva, *INSARJ*

Astrid Moura Vicente, *INSARJ*

Andreia Filipa Neves, *INSARJ*

Acknowledgements

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Welcome

The ***National Health Institute Doutor Ricardo Jorge*** and the ***Organizing Committee*** are pleased to welcome you to the International Workshop ***Biobanking for Health Research*.**

Invited Lectures

Wednesday, September 28th

Conference Opening

Human Biobanking for Public Health

João Lavinha

Department of Genetics, National Health Institute Doutor Ricardo Jorge - Lisbon, Portugal

joao.lavinha@insa.min-saude.pt

Biobanks are increasingly recognised as crucial resources for health research. A biobank can be defined as a collection of biological material and associated [medical, demographic, genetic, environmental, …] data and information stored in an organised system, for a population or a large subset of a population (OECD 2007). Biobanks are set-up to fulfill a wide spectrum of purposes, namely understanding gene x environment/lifestyle interactions, unravelling the molecular basis of disease subtypes and enabling personalized medicine, developing biomarkers, identifying new targets for therapy, and reducing attrition in the discovery and development of novel health interventions.

The situation of public research biobanking in Europe has been the object of a recent survey (Zika et al 2010). Particularly instructive results will be presented concerning a number of issues, including the reason for establishing a biobank, type of data collected and stored, staff involved in the collection of samples, access to banked items (specimens, data) and ethical, legal and social aspects of biobanking. Options and challenges for networking and harmonisation will be discussed in terms of *inter alia* biobank quality, interoperability and sustainability, or the lack of uniform requirements and/or practices regarding, e.g., data and sample sharing between different countries, privacy, feedback of information to donors.

Ongoing, representative transnational population-based biobanks will be reviewed and a selection of large-scale epidemiological studies on genetic and environmental causes of common public health problems (cancer, stroke, chronic noncommunicable diseases and ageing) will be described in more detail.

To conclude, the concept and the enterprise of Public health genomics – as the responsible and effective translation of genome-based knowledge and technology for the benefit of population health – will be evoked.

e-Epidemiology - Adapting Epidemiology Methods for the 21th Century

Jan-Eric Litton

Karolinska Institute, Sweeden

We need better understanding of natural variation in biological parameters and lifestyle factors among healthy individuals, and the identification of the time, age and geographic dynamics of processes that lead to morbidity and eventually to clinically manifest disease. It is only against the background of normality that paths leading to disease can be identified and addressed.

For the first time in medical history the technical prerequisites exist for merging cascades of molecular information from biological samples with demographic and lifestyle data on healthy individuals, and to relate these data to information from administrative registers and to clinical data from hospital databases. The challenge lies in creating a research infrastructure that utilizes modern technology for high throughput genomic data coupled with modern communication technology and it-technology to assembly and organize demographic and lifestyle information. The new infrastructure allows a wealth of new and important research questions to be addressed.

The biobank of National Institute for Health and Welfare, Finland

Markus Perola

National Institute for Health and Welfare, Finland

To guarantee the top level of expertise in modern genetic and biological analyses, we have built an infrastructure that facilitates the collection of genome-wide information on the genetic background of diseases as well as functional information on the molecules that are critical in the disease process. Furthermore we have established the necessary storage, database and computational resources for the expert analyses of the massive amount of collected biological information. Our scientific expertise, technology platforms and large nationwide sample collections facilitate a highly competitive environment for research and education in molecular medicine of the 21st century.

The biobanking wet lab effort is concentrated to Biomedicum Large scale DNA extraction and storage facility. The facility presently houses DNA from about 200 000 individuals and is co-ordinated by National Institute for Health and Welfare. It is equipped with state of the art bar coding system for sample tracking, an automated DNA extraction equipment, liquid handling robots, storage facilities, and tailor made data management tools for optimal confidentiality and quality control.

We are also actively collaborating with the Estonian Genome Project, which has recruited 51515 Estonian voluntary participants for a population-based follow-up cohort.

These and other cohorts will participate in upcoming FP7 application "Large Prospective Cohorts" where we aim to enhance the accessibility of European sample collections to scientists and facilitate networking of European biobank scientists.

The experience of the National DNA Bank Carlos III (Spain)

Andrés García-Montero

National Health Institute Carlos III, Spain

The National DNA Bank Carlos III (BNADN) is an science and technological platform of the University of Salamanca and the Ministry for Science and Innovation whose main goal is to support genetic/genomic research by providing high-quality human samples (DNA, RNA, plasma, cells, tissues) and associated data for individual researchers, research networks, consortia and institutions, assuring its rational and effective, ethical, legal and scientific use.

Since the creation of BNADN in March 2004 till now, efforts have been made to transform it into a unique platform that offers high-quality services and advise on biobanking. Accordingly in this period, apart from building the currently used laboratory facilities and an advanced legal and ethical framework of action, in the BNADN more than 32,000 samples from an identical number of individuals have been collected. Its structure is formed by five different nodes since 2006 (central coordinating node plus four nodes on prevalent diseases: cardiovascular, neuropsychiatric, metabolic and oncological diseases). The current nodal structure involves signed collaborations with more than sixty different institutions, including regional blood transfusion centres, hospital and research centres, private foundations and major national scientific medical associations. The collected samples are structured into healthy controls (n= 5,032; including unrelated donors, trios, twins and 110 individuals with >90 years), population-based cohorts (n=4,382), and patients with oncological (n=3,552), neuropsychiatric (n=1,194), metabolic (n=3,204), cardiovascular (n=1,856), immunologically mediated chronic inflammatory disorders (IMID) (n=10,751), as well as other rheumatologic disorders (n=2,526) and some rare diseases (n=392). From the patients and healthy control sample collections more than 47.000 sample aliquots have already been distributed for more than 80 research projects. In addition, BNADN has also provided other services consisting of automated DNA extraction, cell immortalization education and advisory expertise on biobanking to 22 institutions. In this regard, during the last 6 years >160 professionals have visited the BNADN for training with a mean duration of 3 days/person.

Currently, a total of 15 people work at the BNADN with a strong expertise in biobanking (clinical samples and technological development), advanced flow cytometry (e.g. sorting of pure cell populations and development of new technology and markers for immunophenotyping) and extensive experience in collaboration and/or coordination of National and European networks. Their main research activities focus on biobanking technology development (integrated biobank software design; high throughput cell purification; sample preparation and preservation for -omics; quality control …). For these aims, robotic technologies for nucleic acid extraction and liquid sample handling are available together with a biosecurity-safe level III laboratory facility and high throughput cell separation facility (Flow Cytometry and MACS cell sorters). In turn, equipment and technology for different sample storage formats at room temperature, 4ºC, -80ºC and -196ºC are available. Finally, a proprietary LIMS has been developed which handles all information from the moment a sample/data is donated at a collaborating centre till it is distributed and used by an external research group. To assess the quality of stored DNA samples different technologies are used to control purity, integrity and traceability of DNA samples (electrophoresis/ pulse field electrophoresis, spectrophotometry, fluorimetry, STRs and multiplex and long-expand PCR). Activities of the BNADN has been certified from February 2006 with and ISO 9001 certification.

In turn the activities of the BNADN have been evaluated with recommendations being made by the Ministry of Science and Innovation, the Genoma España Foundation, the University of Salamanca, the Healthcare Department of the Regional Government of Castilla y León, in addition to the two external scientific and ethical committees. Both committees have also evaluated all requests for using the resources of the BNADN, their recommendations being systematically followed.

Portuguese Neonatal Screening Programme Repository

Laura Vilarinho and Hugo Rocha

Newborn Screening Unit, Genetics Department, National Health Institute Doutor Ricardo Jorge (INSARJ), Portugal

Newborn Screening Programs are highly successful public health programs where with simple blood tests can identify neonates at risk for a number of health conditions that if caught early can benefit from early intervention.

The Portuguese Neonatal Screening Programme, funded by the Health Minister, started in 1979 with the screening for phenylketonuria (PKU) and shortly after, congenital hypothyroidism screening (CH) has also been included.

In 2004, with the aim of expanding the number of diseases for which Portuguese newborns are screened for, tandem mass technology was introduced in the National Laboratory allowing the screening for 24 metabolic disorders. Blood spot samples were collected between day 3 and 6 in Whatman 903 filter paper, and newborns screened for a total of 25 disorders.

The Programme is not mandatory and has 99.8% coverage of the country (including Madeira and the Azores islands). The screening of all Portuguese newborns is performed in one single lab that processes over than 400 samples by day, being the decisions on screening centralized.

After newborn screening is complete, a small amount of dried blood remains on the filter paper card. These residual samples (residual blood spots) are stored for a 15 year period, being protected by national regulation (law nº 12/2005). During this time they can be useful to laboratory quality control and assurance monitoring, diagnostic proposes of the newborn (used after written consent from the parents), as well public health and biomedical research (anonymously).

Sometimes, additional analyses are needed for performing the diagnosis of other disorders or if the neonate has deceased with a severe genetic disease without a precise diagnosis. In the later situation and if a new pregnancy occurs, the parents can ask for the residual blood spot sample, so that additional studies can be done to identify the fatal disease. With this information, genetic counselling will be given to the couple as well the possibility of a

prenatal diagnosis. Throughout these years thousands of samples have been of exceptional importance for diagnosis of several genetic and infectious diseases.

Additionally, the residual blood spot samples have been used, anonymously, for research purposes, namely for epidemiological studies, such as HIV/AIDS, hemoglobinopathies and prenatal cytomegalovirus infection in our population.

The use of residual blood spots for diagnosis and research is a well-established side benefit of newborn screening programs.

Thursday, September 29th

Disease and Tissue Biobanks

Building a network of tumour banks in Portugal

Fátima Carneiro

Institute of Molecular Pathology and Immunology (IPATIMUP) and Faculty of Medicine/Hospital S.João, Porto, Portugal

Among biobanking initiatives, Tumour Banks play a pivotal role in biomedical research. The general aim of a Tumour Bank is to acquire neoplastic and control non-neoplastic samples, in standardized conditions for research (basic, clinical or translational). A Tumour Bank is a vital new resource for cancer research, providing high quality, well-characterized tissue.

It is possible for pathologists to collect fresh tissue prospectively during their routine dissection procedures. In this way, the specimens can be optimally sampled and stored for both diagnosis and research purposes. Ideally, specimens are sampled immediately after surgery, prior to fixation, to ensure optimal preservation of proteins and nucleic acids. Retrospective collection of tumour tissue for study and banking purposes is feasible also because, in most countries, pathology laboratories have been legally obliged to file, for at least some years, the formalin-fixed and paraffin-embedded samples that were analyzed.

Over the last decade, Tumour banks acquired a pivotal role in translational research in the field of oncology, providing tools for: evaluation of new predictive factors; evaluation of the value of a known target in a new entity; search for new therapeutic targets; validation of new diagnostic markers; implementation of new diagnostic procedures, namely development of tissue-based diagnostic tests for guidance of therapy with new drugs introduced in clinical practice.

In this scenario, it is a priority to emphasize the central role that pathologists play in translational research, specifically in tumor banking, by the establishment of a bridge between clinicians and basic researchers.

In this presentation it will be presented the steps to establish the Tumour Bank of Hospital S.João[1], as well as the initiatives to build a National Network of Tumour Banks in Portugal [2].

[1] Rodrigues M, Vitó I, Santos R, Paiva J, Pontes P, Silva P, Carneiro F: Establishment of a Tumour Bank: The experience of the Department of Pathology of Hospital S.João (Porto, Portugal). Cell Tissue Bank 10:75–77, 2009. doi: 10.1007/s10561-008-9102-3.
[2] http://www.acs.min-saude.pt/2009/12/18/projectornbt/

Biobanking for discovery of molecular markers in Amyotrophic Lateral Sclerosis: Lisbon's experience

Júlia Costa[1], Mamede de Carvalho[2,3]

[1]*Instituto de Tecnologia Química e Biológica, 2780-157 Oeiras*

[2] *Department of Neurosciences, Hospital de Santa Maria, Lisbon, Portugal*

[3]*Neuromuscular Unit, Instituto de Medicina Molecular, Faculdade de Medicina de Lisboa, Lisbon, Portugal*

Amyotrophic Lateral Sclerosis (ALS) is a fatal neurodegenerative disease of the motor system. The diagnosis is clinical, but electromyography is essential to support diagnosis. Other ancillary investigations, such as transcranial magnetic stimulation and neuroimaging have a great potential. Between 5 to 10% of the ALS cases have a positive familial history, up to now eleven genes have been identified as associated with the disease. The mean age of disease onset occurs between 55-60 years. The incidence is about 2-3 and the prevalence 7-8 per 100.000 in Europe.

The neuropathological hallmarks of ALS have been well studied; however, validated neurochemical biomarkers to provide early diagnosis, for monitoring disease progression, to define prognosis and unveil pathophysiological pathways are still missing. At present several candidate biomarkers exist, but a large number of samples are required for independent verification and translation into clinical application.

In Lisbon, in collaboration between Faculty of Medicine of Lisbon and Instituto de Tecnologia Química e Biológica a bank of plasma and cerebrospinal fluid of patients with ALS, controls with other neurological disorders and healthy controls has been initiated. At present the bank has more than 290 plasma samples and 30 cerebrospinal fluid samples.

Indications on standard operating procedures for biological tissue collection and conservation to facilitate multi-centre collaboration, validation and ultimately clinical translation of biomarkers in ALS will be presented.

Results from the biochemical characterization of the samples will be shown.

EuroBioBank: an almost ten year experience in biobanking for rare diseases

Angelini C., Baumeister S. , Ben Yaou R., Bignami F., di Donato J-H., Felice A., Garavaglia B., Gurwitz D., Karcagi V., Lochmüller H., Meznaric M., Moggio M., **Mora M.**, Politano L., Posada M., Saker-Delye S., Schneiderat P., Voit T.

Carlo Besta Neurological Institute, Italy

EuroBioBank, the first operating European Network of DNA, Cell and Tissue Banks for Rare Diseases, was established in 2001 to facilitate access to quality human biomaterials for researchers worldwide.

Main objectives of the project were:

- to improve links between existing Biological Resources Centres, in order to identify and localise the biological material (DNA, tissue, cell cultures) at European level,
- to optimise the use of existing collections and boost the creation of new ones,
- to harmonise and spread quality banking practices in the respect of ethical issues,
- to distribute quality material and associated data to all users gathering and making available the information concerning this material through a dedicated database,
- to disseminate knowledge and know-how to the scientific community.

Composed of 13 biobanks from 7 EU countries and coordinated by EURORDIS (European Organisation for Rare Diseases), the network was originally funded by the European Commission within the European 5th Framework Programme (2003-2006).

The EuroBioBank network was then selected to be the biobanking service structure within the TREAT-NMD Network of Excellence (FP6: 2007-2012) with EURORDIS taking the leadership of WP04.1:“Develop and Manage Supranational BioBanks”. As such, it received EC funding for coordination of the network and hosting of the website, while each biobank of the network is financed by its own national institution or charitable organisation. Moreover, a collaboration has been established with BBMRI (European Biobanking & Biomolecular Resources Research Infrastructure).

A main objective of the EuroBioBank Network is to continue improving the availability, exchange and use of human biomaterials for translational research on neuromuscular disorders. A central tool to reach this objective is the EuroBioBank website www.eurobiobank.org which

provides services for TREAT-NMD researchers, such as the online catalogue of samples, Standard Operating Procedures (SOPs) and ethical documents. Implementation of quality control for biomaterials from NMD patients and additional evaluation of the SOPs are also part of this project.

The EuroBioBank Network is currently composed of 18 members, of which 16 biobanks from 8 European countries (France, Germany, Hungary, Italy, Malta, Slovenia, Spain and the United-Kingdom) as well as Israel and Canada.

Over 440000 samples are available across the EuroBioBank Network. Approximately 13000 samples are collected each year and 7000 distributed to researchers, and more than 130 peer-reviewed studies have been published acknowledging EuroBioBank.

By facilitating access to high quality DNA, cell and tissue samples for the worldwide research community, while working closely with Research Ethics Committees (RECs) and maintaining ethical standards along donors'consent, the EuroBioBank Network thus plays a major role in the acceleration of cutting edge treatments for rare diseases.

IT infrastructure for biobanking

Demiroglu, S.Y.[1]

[1] *Georg-August-University Goettingen, University Medical Center, Dept. of Medical Informatics, Robert-Koch-Str. 40, 37075 Goettingen, Germany*

From an IT perspective, biobanks consist of more than a freezer filled with biomaterial and the data on the storage location. They form the core component of a whole research infrastructure. Biomaterial on its own is of little value, but it yields value with the amount of data that is linked to it.

Therefore, an IT infrastructure for biobanking consists of several components, which map the handling processes from patient inclusion to requesting biomaterial samples. The components provide functionalities for human subject management, subjects' identity management, and biomaterial management, as well as an electronic lab book, a phenotype database, an medical image database, an analysis database, and a research database.

A human subject management tool administrates the rounds of participants, their contact details, and the status of the informed consents, and additionally holds a description of genetic relationships between the participants. The identity of the subjects is managed at the subjects' identity management, which administrates pseudonyms and identifying data and should be located at a trusted third party to ensure data privacy. The biomaterial management should contain the type, quality, and storage location of each biological sample. In an electronic lab book, all processes which led either to the extraction of biomaterial or the acquisition of analysis data should be described in detail. In the phenotype database clinical and medical data of the human subjects are stored, this could also include data acquired by questionnaires. Pictures should be stored in a specialized image database, preferably a PACS (picture archiving and communication system). An analysis database contains all analysis data gained from the biomaterial, e.g. genetic sequences. Finally, in a research database the data from the databases and biomaterial management described above can be (dynamically) linked together pertaining to the research question. Furthermore, requests for biomaterial can be handled using the research database because in this view phenotypes and analysis data are linked to the biomaterial.

To avoid entering the same data into multiple systems interfaces from routine health care to the biobank infrastructure should also be implemented. Here, the de-identification of patient-

related data and the fact that the documentation is mostly done for cost-reimbursement, and thus holds little usable information, are the biggest challenges.

Friday, September 30th

Laboratory Sample Management

Considerations in the management of the finite UK Biobank sample resource to maximise its scientific research utility

Paul Downey MBA

Operations Director

UK Biobank Units 1-3 Spectrum Way, Adswood
Stockport, Cheshire, SK3 0SA
Web: www.ukbiobank.ac.uk

UK Biobank* is a prospective epidemiological project; its aim is to provide a valuable research resource to the biomedical research community, globally.

The 500,000 UK wide volunteers who participated in the recruitment phase (2007-2010) donated samples of: blood, urine and saliva. These primary samples were processed further, resulting in 13.3 million two dimensionally barcode labelled sample aliquots, that are stored in a bespoke automated -80^{0}C store and a subset stored as a backup in vapour phase liquid nitrogen.

UK Biobank will invite applications from researchers interested in using the resource from Q3 2011. The talk will describe the challenges and approaches to creating and managing the resource as well as the strategies to maximise the scientific return from the finite sample collection.

*http://www.ukbiobank.ac.uk/

An NCI Perspective on Creating Sustainable Biospecimen Resources

Jim Vaught, Ph.D.

Deputy Director
Office of Biorepositories and Biospecimen Research
http://biospecimens.cancer.gov/

National Cancer Institute, USA 11400 Rockville Pike Bethesda, MD 20892
phone 301-451-7314

High quality biospecimens with appropriate clinical annotation are critical in the era of personalized medicine. It is now widely recognized that biospecimen resources need to be developed and operated under established scientific, technical, business, and ethical/legal standards. To date such standards have not been widely practiced, resulting in variable biospecimen quality that may compromise research efforts.

The U.S. National Cancer Institute (NCI) Office of Biorepositories and Biospecimen Research (OBBR) was established in 2005 to coordinate NCI's biospecimen resource activities and address those issues that affect access to the high quality specimens and data necessary for its research enterprises as well as the broader translational research field. OBBR and the NCI Biorepository Coordinating Committee developed NCI's Best Practices for Biospecimen Resources after consultation with a broad array of experts. A Biospecimen Research Network was established to fund research to develop additional evidence-based practices. While these initiatives will improve the overall availability of high quality specimens and data for cancer research, OBBR has been authorized to implement a national biobanking effort, caHUB (cancer HUman Biobank). caHUB will be a unique public resource that contributes to scientific and medical advances by providing high quality, fully annotated human biospecimens and associated services to the research and product development communities.

This presentation will outline the progressive efforts by NCI in various technical, governance, and economic considerations that will be important as the new caHUB enterprise is undertaken.

Anne Cambon-Thomsen
INSERM, France

Confidentiality and data protection issues, the new paradigm

Paula Lobato de Faria

Escola Nacional de Saúde Pública – Universidade Nova de Lisboa

Lisbon/Portugal

Confidentiality has become an almost trivialised term. This trivialisation is evident in a range of activities, from the academic debates in bioethics and biolaw to the professional level, encompassing the idosyncratic practices of healthcare and scientific research units. When genetic data is the subject, this trivialisation is taken to a yet unprecedented level, being used and abused as if it possesed supernatural powers, capable of transmuting anything into an ethically and legally acceptable fact. Nevertheless, in legal terms, the right to confidentiality is a very recent phenomenon, and although it is rooted in the right to privacy as well as the right to informational self-determination, it is neither identifiable with them, nor does it constitute an independent legal figure in the panorama of fundamental rights, the contours of which, are probably less known than they appear to be.

Hence, in order to effectively talk about confidentiality in biobanks, we first need to know both the origins and the present juridical characterization of the "right to confidentiality" in Health Law, which includes the analysis of its *ratio*, legal sources, object, contents, recipients, and limits. Mapping confidentiality in the Health Law territory allows for a deeper understanding of the real power of this often overestimated concept in the legal and ethics debates on research biobanking.

It is a well-known fact that the effectiveness of fundamental human rights is constrained, not only by inherent legal insufficiencies (*e.g.* how to internationally sanction corrupt governments?), but also by social, cultural and economic factors that engage in the undermining of the legislator's goal, in the complex process of creating a new human right. These "limiting factors" play a significant role in what relates to the right to confidentiality, either in healthcare in general or in biobanks in particular. For this reason, it is reasonable to ask whether confidentiality and data protection have become nothing more than paper rights?

Amongst the mentioned factors that limit the real existence of a right to confidentiality and data protection in research biobanking, it is helpful to distinguish between extrinsic legal insufficiencies and factors that are extrinsic to the legal dimension: The former comprise difficulties pertaining to efficient sanctions due to imprecisions relating to the object of the right.

The latter include, but is not limited to, the impossibility of totally preventing *hacker* attacks on the biobank databases and the growing sense of public health genetics that implicitly involves the establishment of a common right to "genetic information for all", which is the negative correlate of "confidential genetics".

In light of the previous reflexion, we argue that presenting confidentiality and data protection as fundamental legal pillars of ethically conscious research biobanking may, at present, not only be fallacious, but also obsolete, as these rights need a profound legal reflexion based on the new paradigms of our current society.

Participants

Survey on existing biological sample repositories in INSA

Ribeiro, J.[1] and Santos, R.[2]

[1]*Cytogenetics Unit, Centro de Genética Médica Dr. Jacinto Magalhães, Instituto Nacional de Saúde Dr. Ricardo Jorge I.P., Porto, Portugal*

[2]*Molecular Genetics Unit, Centro de Genética Médica Dr. Jacinto Magalhães, Instituto Nacional de Saúde Dr. Ricardo Jorge I.P., Porto, Portugal*

Knowledge of disease mechanisms and development of translational research requires access to well-characterized biological samples. Biobanks, defined as repositories of samples and associated data stored in a systematic and standardized fashion under controlled conditions, have emerged as a crucial source to generate new opportunities for research and development.

The National Institute of Health Dr. Ricardo Jorge (INSA) harbours a vast number of unique sample collections, obtained via research or diagnostic activity, with enormous potential for biomedical research. Some collections are representative of the general population or of specific subpopulations, and thus useful epidemiological studies and determination of prevalence rates, or in the study of interactions between genes, lifestyle and social and environmental factors; others are disease-specific, such as cardiovascular disease, blood disorders or mental retardation. There are also collections of samples in different areas of study which are of great value as reference material.

In order to determine the true potential of existing collections and to plan the implementation of a biobank at INSA, a general survey was carried out across all Departments. The questionnaire aimed to assess:

- nature of collections stored, assessing its size, purpose and importance;

- available infrastructures (physical location, equipment and monitoring conditions);

- nature of associated data (type of information, identification and records);

- existence and type of associated informed consent;

- mechanisms in place to ensure confidentiality, privacy and access to samples and associated data;

- quality control procedures.

This survey identified 13 main areas of study, where samples have been stored for diverse purposes, including epidemiological studies, specific research projects, research applied to health care as well as for technological development.

We present the scope, approximate size and conditions of the collections reported. This information constitutes as a starting point for stakeholders to evaluate crucial aspects regarding the implementation of a biobank at INSA, namely the strategic importance and potential of existing repositories, the necessary infrastructures, the aim and scope of the biobank and its management and sustainability.

Facts related to the collection of biological samples in the National Health Examination Survey - Portuguese Component of the European Health Examination Survey

M. Barreto[1], V. Francisco[1], P.Rasteiro[2], E. Sousa[2], A. Vicente[1], M. Bourbon[1], A.Fernandes[2], A. Beleza[2], F. Mendonça[3], A. Gil[1], C. Matias Dias[1].

[1]Instituto Nacional de Saúde Doutor Ricardo Jorge, Lisboa, Portugal; [2]Laboratório de Saúde Pública Dra. Laura Ayres, Faro, Portugal; [3]Administração Regional de saúde do Algarve, Faro, Portugal.

The objective of the National Health Examination Survey (NHES), which corresponds to the Portuguese component of the European Health Examination Survey (EHES), is to collect health data, related risk factors and biological samples of the Portuguese population, using the EHES recommended methodology. These surveys involve an interview, clinical and physical measurements and blood collection. In this context, we herein describe the pilot study performed in S. Brás de Alportel in the Algarve region. For this pilot study, we have recruited 221 individuals (95 males and 126 females), between 25 and 91 years old, who were enrolled in the Health Centre of S. Brás de Alportel (Algarve). For each participant, we have collected 16.5 ml of total blood, in five different Vacutainer® tubes, which was later processed into serum, plasma and DNA. We have performed several biochemical analyses(total cholesterol, LDL,HDL, glucose, tryglicerides, creatinine, ALT, AST, _-GT, CRP and iron) and a complete blood count. From the 221 participants in this pilot study, we were

able to collect blood to 219 (99.5%). To 185 of these (84.5%) we were able to collect the total amount of blood. The biochemical analyses were performed in all the samples. The total blood count was performed in 103 samples (47%) due to transport constraints. We have also collected DNA from 210 participants (95.9%). We have created a biobank comprising 1847 serum aliquots and 959 plasma aliquots, which have been stored at -80°C and 210 DNA aliquots which have been stored at 4°C. In conclusion, during this study, we have optimized the logistics and procedures to perform the large scale study for the NHES and EHES. In addition, we have created a biobank comprising detailed questionnaire data, physical and clinical data and biological samples from a representative sample of S. Brás de Alportel in Algarve, Portugal. This biobank will allow us to perform future studies, including the determination of the prevalence of gene variants of public health interest, the characterization of gene-environment interactions in the development of chronic diseases and the genetic structure of the Portuguese population. The

success rate, the quality of the data and of the biological samples was high and comparable to similar studies.

Understanding health and diseases in the life course – The Porto Population-based biobank

Barros, H.[1, 2], Lopes, C.[1, 2], Lunet, N.[1, 2], Ramos, E.[1, 2], Azevedo, A.[1, 2], Santos, AC.[1, 2], Lucas, R.[1, 2], Peleteiro, B.[1, 2], Fraga, S.[1, 2], Sousa, S.[1, 2], Guimarães, JT.[2]

[1]Department of Clinical Epidemiology, Predictive Medicine and Public Health

University of Porto Medical School

[2]University of Porto Institute of Public Health

The Department of Clinical Epidemiology, Predictive Medicine and Public Health of the University of Porto Medical School, currently coordinates two population-based cohorts [a cohort of adolescents born in 1990 and assembled at 13 years (*EPITeen*) and an adult cohort (*EPIPorto*)]. These cohorts comprise representative samples of uninstitutionalized individuals living in Porto. They focus on genetic, social and environmental health determinants with particular attention to the consequences of psychosocial (such as violence, quality of life, depression) and behavioural (smoking, alcohol consumption, diet and physical activity) characteristics. Major diseases such as cardiovascular, musculoskeletal, infectious and oncologic are addressed and the role of different levels of exposure and causal paths are tested to provide ethiological clues.

Participants from all the cohorts provided biological samples that are stored at -20 ºC and/or -80 ºC in the biobank. This biobank, currently comprises around 64000 biological (plasma, serum) samples, collected at different longitudinal time points, from the 5000 participants. Both EPIPorto and EPITeen stored, at -80ºC, samples of washed erythrocytes, after plasma separation in k3EDTA-treated tubes, offering DNA. The variety of biological samples stored, their longitudinal collection, coupled with an extensive amount of information obtained through questionnaires, physical exams and medical records will provide answers to numerous scientific questions and essential information for adequate planning and management of preventive measures. The biobank and the linked information has been used also for academic purposes and were the material base for eight PhD dissertations, 50 Master thesis and more than 100 international papers.

The Porto birth cohort biobank

Barros, H.[1, 2], Correia, S.[1, 2], Guimarães, JT.[2], Santos, AC.[1, 2]

[1]Department of Clinical Epidemiology, Predictive Medicine and Public Health
University of Porto Medical School
[2]University of Porto Institute of Public Health

Geração XXI is the first prospective population-based birth cohort assembled in Portugal. Geração XXI has been conducted in Porto Metropolitan Area, in the North of Portugal. The baseline assembling comprised 8647 newborns whose mothers were invited to participate in the study. The recruitment occurred at five level III maternity units in the Porto metropolitan, between April 2005 and August 2006. Several data sources were used such as structured questionnaires for the mother and the father, the mother and newborn medical registries and anthropometry. Venous blood samples were collected from the umbilical cord, the mother and, whenever possible, the father. These samples were immediately processed and stored. The Geração XXI biobank also comprises a paper dried blood spots from each child, collected at time of birth as part of the Early Diagnosis National Program.

Four years after birth, the first evaluation of the entire cohort was performed and for a subsample of 2754 mother and 1526 children, blood samples were collected and stored.
All the procedures were explained to the participants and a written informed consent is always evaluated.

The Porto birth cohort biobank is a population-based biobank with a comprehensive collection of genetic, phenotypic and environmental information. It comprises different biological samples, such as serum, plasma, umbilical cord blood, and whole blood, stored at -80^{o}C.

Currently, the Geração XXI biobank contains about 50 000 serum samples, 13 000 plasma, 1700 umbilical cord blood, and whole blood samples from both the mother (n= 900) and father (n=500) of the Geração XXI children.

This biobank is an infrastructure for research that together with the availability of extensive questionnaire data and serial measurements provides an impressive source of

information on Portuguese child's health determinants and its influence on health-related outcomes during the lifecourse.